

Supplemental material to Luise Hartmann, Judith S. Hecker, et al.:

Compartment-specific mutational landscape of clonal hematopoiesis

Supplemental Methods

Flow-sorting and genetic analyses of mature and precursor cell populations from BM samples

Antibodies used for cell sorting are listed in Supplemental Table 1.

After thawing BM samples, one fourth of the sample was stained with 100µl of an antibody mix for sorting mature cell populations (granulocytes, T lymphocytes, and B lymphocytes) along with CD34⁺ cells (Supplemental Figure 1A). Dead cell exclusion was performed using DAPI 1µg/ml (BioLegend, #422801). The rest of the sample was sorted into fractions containing hematopoietic stem cells (HSC: lin⁻CD34⁺CD38⁻CD45RA⁻CD90⁺), common-myeloid progenitors (CMP: lin⁻CD34⁺CD38⁺CD45RA⁻CD123⁺), granulocyte-monocyte progenitors (GMP: lin⁻CD34⁺CD38⁺CD45RA⁺CD123⁺) and megakaryocyte-erythroid progenitors (MEP: lin⁻CD34⁺CD38⁺CD45RA⁻CD123⁻) (Supplemental Figure 1B). For this panel, dead cells were excluded using propidium iodide 0.4µg/ml (Sigma, #P4170). Sorting was performed using a BD FACS Aria III cell sorter and a 100µm nozzle.

DNA from flow-sorted cells (9.000-200.000 cells per population) was isolated using the QIAamp DNA Micro kit (Qiagen, Hilden, Germany). A custom unique molecular identifier-based QIAseq gene panel (Qiagen) targeting *ASXL1* exon 13 was used for library preparation, and sequencing was performed on a MiSeq instrument (Illumina, San Diego, USA). Variants were detected using the QIAGEN CLC Genomics Workbench. VAFs within flow-sorted cell populations were compared using the Friedman test followed by Dunn's multiple comparison test. *ASXL1* mutation VAFs in each flow-sorted population were compared to the same individual's bulk BM cells using Fisher's test, with multiple testing correction using the Benjamini-Hochberg procedure.

Supplemental Tables

Supplemental Table 1: List of antibodies used for FACS sorting of mature and progenitor BM cell populations.

Panel	Antibody	Conjugate	Dilution	Company	Catalogue Number
Mature	Anti-CD3	APC-Cy7	1:30	BioLegend	344818
Mature	Anti-CD15	FITC	1:10	Beckman Coulter	B36298
Mature	Anti-CD19	PE	1:30	BioLegend	302208
Mature	Anti-CD34	PE-Cy7	1:30	BioLegend	343616
Mature	Anti-CD45	APC	1:100	eBioscience	17-0459-42
Progenitor	Anti-CD4	biotin	1:100	BioLegend	300504
Progenitor	Anti-CD8a	biotin	1:100	BioLegend	301004
Progenitor	Anti-CD15	biotin	1:100	BioLegend	301914
Progenitor	Anti-CD19	biotin	1:100	BioLegend	302204
Progenitor	Anti-CD34	FITC	1:15	BD Pharmingen	555821
Progenitor	Anti-CD38	APC	1:25	BD Biosciences	345807
Progenitor	Anti-CD45	PerCP-Cy5.5	1:100	BioLegend	304028
Progenitor	Anti-CD45RA	PE-Cy7	1:20	BD Biosciences	560675
Progenitor	Anti-CD90	PE	1:25	BD Biosciences	555596
Progenitor	Anti-CD123	BV421	1:25	BioLegend	306018
Progenitor	Anti-CD235a	biotin	1:100	Invitrogen	13-9987-80
Progenitor	Straptavidin	APC-Cy7	1:100	BioLegend	405208

Supplemental Table 2: Variants identified in individuals with clonal hematopoiesis.

Subject	Gene	Variant	Mutation type	VAF [%]	Specimen
UPN_1	ASXL1	NM_015338.6:c.2036dup;p.(Gly680Argfs*38)	InDel	3.71	BM
UPN_1	ASXL1	NM_015338.6:c.2385del;p.(Trp796Glyfs*22)	InDel	5.93	BM
UPN_1	STAT3	NM_139276.2:c.1940A>T;p.(Asn647Ile)	Missense	1.79	BM
UPN_1	TET2	NM_001127208.2:c.5760A>T;p.(Lys1920Asn)	Missense	4.75	BM
UPN_1	U2AF1	NM_001025203.1:c.470A>C p.(Gln157Pro)	Missense	9.73	BM
UPN_10	BCORL1	NM_021946.4:c.4013G>A;p.(Arg1338Gln)	Missense	6.39	BM
UPN_10	RAD21	NM_006265.3:c.65A>G;p.(His22Arg)	Missense	4.49	BM
UPN_100	ZBTB7A	NM_015898.4:c.149C>T;p.(Ser50Leu)	Missense	3.18	BM
UPN_11	BCORL1	NM_021946.4:c.4853+1G>C;p.?	Splice site	10.86	BM
UPN_12	DNMT3A	NM_175629.2:c.2567_2568delAG;p.(Glu856Glyfs*7)	InDel	1.56	PB
UPN_12	TET2	NM_001127208.2:c.3954+5G>A;p.?	Splice site	1.13	PB
UPN_13	DNMT3A	NM_175629.2:c.2393T>C;p.(Leu798Pro)	Missense	1.00	BM
UPN_13	TET2	NM_001127208.2:c.5582G>A;p.(Gly1861Glu)	Missense	4.66	BM
UPN_14	DNMT3A	NM_175629.2:c.2339T>C;p.(Ile780Thr)	Missense	1.14	BM
UPN_14	TET2	NM_001127208.2:c.5686A>G;p.(Arg1896Gly)	Missense	5.34	BM
UPN_15	DNMT3A	NM_175629.2:c.1123-2A>G;p.?	Splice site	23.6	BM
UPN_15	TET2	NM_001127208.2:c.4079T>G;p.(Leu1360Arg)	Missense	1.19	BM
UPN_16	DNMT3A	NM_175629.2:c.2106T>G;p.(Asp702Glu)	Missense	3.59	BM
UPN_16	IDH2	NM_002168.3:c.678+1G>T;p.?	Splice site	1.07	BM
UPN_16	TET2	NM_001127208.2:c.1842dup;p.(Leu615Alafs*23)	InDel	1.18	BM
UPN_17	DNMT3A	NM_175629.2:c.1940T>G;p.(Leu647Arg)	Missense	4.47	BM
UPN_17	TET2	NM_001127208.2:c.4136C>A;p.(Ala1379Asp)	Missense	2.42	BM
UPN_17	TET2	NM_001127208.2:c.3116C>A;p.(Ser1039*)	Nonsense	2.81	BM
UPN_18	DNMT3A	NM_175629.2:c.2092T>G; p.(Trp698Gly)	Missense	5.77	PB
UPN_19	DNMT3A	NM_175629.2:c.2195T>C; p.(Phe732Ser)	Missense	1.15	PB
UPN_2	ASXL1	NM_015338.6:c.2083C>T;p.(Gln695*)	Nonsense	1.29	BM
UPN_2	DNMT3A	NM_175629.2:c.2141C>G;p.(Ser714Cys)	Missense	1.08	BM
UPN_2	JAK2	NM_004972.3:c.1849G>T, p.(Val617Phe)	Missense	5.96	BM
UPN_20	DNMT3A	NM_175629.2:c.2591T>A; p.(Met864Lys)	Missense	21.29	PB
UPN_201	DNMT3A	NM_175629.2:c.776delC;p.(Ala259Valfs*57)	InDel	1.53	BM
UPN_201	DNMT3A	NM_175629.2:c.942G>A;p.(Trp314*)	Nonsense	5.39	BM
UPN_201	TET2	NM_001127208.2:c.5618T>C;p.(Ile1873Thr)	Missense	2.78	BM
UPN_204	TET2	NM_001127208.2:c.3594+5G>T;p.?	Splice site	2.90	BM
UPN_204	TET2	NM_001127208.2:c.4585C>T;p.(Gln1529*)	Nonsense	2.44	BM
UPN_205	DNMT3A	NM_175629.2:c.1506del p.(Thr503Profs*148)	InDel	4.28	BM
UPN_205	NFE2	NM_001136023.3:c.814C>T;p.(Arg272*)	Nonsense	4.31	BM
UPN_206	DNMT3A	NM_175629.2:c.2408+6T>G;p.?	Splice site	1.41	BM
UPN_208	DNMT3A	NM_175629.2:c.2204A>G;p.(Tyr735Cys)	Missense	1.24	BM
UPN_21	DNMT3A	NM_175629.2:c.2047_2056del; p.(Tyr683Thrfs*19)	InDel	3.41	PB
UPN_213	TET2	NM_001127208.2:c.4791_4792del;p.(Phe1597Leufs*16)	InDel	1.39	BM
UPN_213	TET2	NM_001127208.2:c.444_447del;p.(Lys148Asnfs*3)	InDel	2.12	BM
UPN_214	PPM1D	NM_003620.4:c.1654C>T;p.(Arg552*)	Nonsense	10.63	BM

Subject	Gene	Variant	Mutation type	VAF [%]	Specimen
UPN_216	<i>PPM1D</i>	NM_003620.4:c.1654C>T;p.(Arg552*)	Nonsense	2.00	BM
UPN_218	<i>DNMT3A</i>	NM_175629.2:c.2086del;p.(Gln696Argfs*9)	InDel	1.16	BM
UPN_22	<i>DNMT3A</i>	NM_175629.2:c.2711C>T;p.(Pro904Leu)	Missense	6.64	BM
UPN_220	<i>DNMT3A</i>	NM_175629.2:c.2711C>T;p.(Pro904Leu)	Missense	1.16	BM
UPN_222	<i>TET2</i>	NM_001127208.2:c.2340_2343del;p.(Val781Lysfs*31)	InDel	1.77	BM
UPN_223	<i>DNMT3A</i>	NM_175629.2: c.2645G>A p.(Arg882His)	Missense	3.04	BM
UPN_225	<i>DNMT3A</i>	NM_175629.2:c.1674T>G;p.(Phe558Leu)	Missense	1.28	BM
UPN_225	<i>TET2</i>	NM_001127208.2:c.1441C>T;p.(Gln481*)	Nonsense	1.30	BM
UPN_226	<i>ASXL1</i>	NM_015338.6:c.2179G>T;p.(Glu727*)	Nonsense	2.67	BM
UPN_227	<i>PPM1D</i>	NM_003620.4:c.1709C>G;p.(Ser570*)	Nonsense	1.18	BM
UPN_23	<i>DNMT3A</i>	NM_175629.2:c.1506delT; p.(Thr503Profs*148)	InDel	4.73	PB
UPN_231	<i>DNMT3A</i>	NM_175629.2:c.2339T>C;p.(Ile780Thr)	Missense	1.64	BM
UPN_231	<i>DNMT3A</i>	NM_175629.2:c.2408G>A;p.(Arg803Lys)	Missense	1.79	BM
UPN_231	<i>DNMT3A</i>	NM_175629.2:c.842_855del;p.(Glu281Glyfs*38)	InDel	3.98	BM
UPN_232	<i>ASXL1</i>	NM_015338.6:c.1934dup;p.(Gly646Trpfs*12)	InDel	1.20	BM
UPN_232	<i>DNMT3A</i>	NM_175629.2:c.1603T>C;p.(Ser535Pro)	Missense	2.88	BM
UPN_232	<i>TET2</i>	NM_001127208.2:c.3765C>A;p.(Tyr1255*)	Nonsense	1.45	BM
UPN_233	<i>DNMT3A</i>	NM_175629.2:c.2207G>A;p.(Arg736His)	Missense	6.75	BM
UPN_235	<i>U2AF1</i>	NM_001025203.1:c.470A>G p.(Gln157Arg)	Missense	2.07	BM
UPN_235	<i>WT1</i>	NM_001198551.1:c.473G>A;p.(Arg158His)	Missense	2.86	BM
UPN_238	<i>DNMT3A</i>	NM_175629.2:c.1123-1G>C;p.?	Splice site	8.24	BM
UPN_24	<i>DNMT3A</i>	NM_175629.2:c.1258A>T;p.(Lys420*)	Nonsense	3.99	BM
UPN_242	<i>ASXL1</i>	NM_015338.6:c.2885_2886del;p.(Val962Alafs*7)	InDel	1.87	BM
UPN_243	<i>TET2</i>	NM_001127208.2:c.2919C>A;p.(Cys973*)	Nonsense	1.89	BM
UPN_245	<i>TP53</i>	NM_000546.5:c.623A>G;p.(Asp208Gly)	Missense	1.35	BM
UPN_248	<i>ASXL1</i>	NM_015338.6:c.2258_2259dup;p.(Thr754Profs*19)	InDel	4.87	BM
UPN_248	<i>SH2B3</i>	NM_005475.2:c.1522dup;p.(Arg508Profs*38)	InDel	40.42	BM
UPN_248	<i>TET2</i>	NM_001127208.2:c.5165del;p.(Pro1722Leufs*23)	InDel	30.10	BM
UPN_248	<i>TET2</i>	NM_001127208.2:c.5481_5482del;p.(Gln1828Alafs*17)	InDel	32.3	BM
UPN_25	<i>DNMT3A</i>	NM_175629.2:c.994G>A;p.(Gly332Arg)	Missense	2.30	BM
UPN_250	<i>TET2</i>	NM_001127208.2:c.2375C>A;p.(Ser792*)	Nonsense	1.10	BM
UPN_251	<i>TET2</i>	NM_001127208.2:c.5618T>C;p.(Ile1873Thr)	Missense	1.15	BM
UPN_251	<i>DNMT3A</i>	NM_175629.2:c.2530A>G;p.(Lys844Glu)	Missense	1.45	BM
UPN_252	<i>SF3B1</i>	NM_012433.3c.1986C>G;p.(His662Gln)	Missense	1.42	BM
UPN_252	<i>TET2</i>	NM_001127208.2:c.5686A>T;p.(Arg1896Trp)	Missense	2.10	BM
UPN_254	<i>SRSF2</i>	NM_003016.4:c.242A>T;p.(Asp81Val)	Missense	3.20	BM
UPN_254	<i>TET2</i>	NM_001127208.2:c.5650A>G;p.(Thr1884Ala)	Missense	43.88	BM
UPN_255	<i>PIGA</i>	NM_002641.3:c.217G>A;p.(Ala73Thr)	Missense	4.95	BM
UPN_26	<i>DNMT3A</i>	NM_175629.2:c.2332G>A;p.(Val778Met)	Missense	5.07	BM
UPN_266	<i>DNMT3A</i>	NM_175629.2:c.2207G>A;p.(Arg736His)	Missense	7.17	BM
UPN_267	<i>DNMT3A</i>	NM_175629.2:c.1238del;p.(Gly413Alafs*238)	InDel	1.00	BM
UPN_27	<i>DNMT3A</i>	NM_175629.2:c.1271del;p.(Pro424Hisfs*227)	InDel	1.49	BM
UPN_272	<i>SRP72</i>	NM_006947.3:c.869A>G;p.(Asn290Ser)	Missense	3.16	PB
UPN_275	<i>DNMT3A</i>	NM_175629.2:c.2657del;p.(Gln886Argfs*20)	InDel	1.94	PB

Subject	Gene	Variant	Mutation type	VAF [%]	Specimen
UPN_276	<i>DNMT3A</i>	NM_175629.2:c.1238dup;p.(Phe414Leufs*7)	InDel	1.42	PB
UPN_28	<i>DNMT3A</i>	NM_175629.2:c.2711C>T;p.(Pro904Leu)	Missense	5.33	BM
UPN_280	<i>PPM1D</i>	NM_003620.4:c.1332_1335del;p.(Phe445Glnfs*5)	InDel	1.24	PB
UPN_282	<i>DNMT3A</i>	NM_175629.2:c.2190_2215del;p.(Phe731*)	InDel	10.81	PB
UPN_283	<i>DNMT3A</i>	NM_175629.2:c.2207G>A;p.(Arg736His)	Missense	5.68	PB
UPN_283	<i>SF3B1</i>	NM_012433.3:c.1997A>G p.(Lys666Arg)	Missense	2.69	PB
UPN_288	<i>DNMT3A</i>	NM_175629.2:c.1903C>T;p.(Arg635Trp)	Missense	1.45	PB
UPN_288	<i>DNMT3A</i>	NM_175629.2:c.2086C>T;p.(Gln696*)	Nonsense	2.60	PB
UPN_29	<i>DNMT3A</i>	NM_175629.2:c.2322+2T>C;p.?	Splice site	1.35	PB
UPN_291	<i>DNMT3A</i>	NM_175629.2:c.2245C>T;p.(Arg749Cys)	Missense	1.64	PB
UPN_291	<i>DNMT3A</i>	NM_175629.2:c.1851+1G>A;p.?	Splice site	2.03	PB
UPN_3	<i>ASXL1</i>	NM_015338.6:c.2954_2957del;p.(Ile985Thrfs*7)	InDel	4.73	BM
UPN_30	<i>DNMT3A</i>	NM_175629.2:c.1948C>G;p.(Leu650Val)	Missense	1.06	PB
UPN_31	<i>DNMT3A</i>	NM_175629.2:c.1904G>A;p.(Arg635Gln)	Missense	6.54	PB
UPN_32	<i>DNMT3A</i>	NM_175629.2:c.2204A>G;p.(Tyr735Cys)	Missense	2.65	PB
UPN_33	<i>DNMT3A</i>	NM_175629.2:c.2330C>G;p.(Pro777Arg)	Missense	1.32	BM
UPN_34	<i>DNMT3A</i>	NM_175629.2:c.2393T>A, p.(Leu798His)	Missense	4.37	BM
UPN_35	<i>DNMT3A</i>	NM_175629.2:c.2401A>G; p.(Met801Val)	Missense	12.06	BM
UPN_36	<i>DNMT3A</i>	NM_175629.2:c.1015-2A>G;p.?	Splice site	4.76	BM
UPN_37	<i>DNMT3A</i>	NM_175629.2:c.1532G>A;p.(Gly511Glu)	Missense	1.49	BM
UPN_38	<i>DNMT3A</i>	NM_175629.2:c.2185C>T;p.(Arg729Trp)	Missense	8.48	BM
UPN_39	<i>DNMT3A</i>	NM_175629.2:c.2644C>T;p.(Arg882Cys)	Missense	1.06	BM
UPN_4	<i>ASXL1</i>	NM_015338.5:c.2512_2537dup; p.(Ser846Argfs*5)	InDel	11.42	PB
UPN_4	<i>ASXL1</i>	NM_015338.6:c.1934dup;p.(Gly646Trpfs*12)	InDel	6.07	BM
UPN_40	<i>DNMT3A</i>	NM_175629.2:c.1900A>T; p.(Ile634Phe)	Missense	1.07	BM
UPN_41	<i>DNMT3A</i>	NM_175629.2:c.1430-3C>G;p.?	Splice site	3.08	BM
UPN_42	<i>DNMT3A</i>	NM_175629.2:c.2377T>C;p.(Tyr793His)	Missense	1.74	BM
UPN_43	<i>DNMT3A</i>	NM_175629.2:c.2141C>G;p.(Ser714Cys)	Missense	2.14	BM
UPN_44	<i>DNMT3A</i>	NM_175629.2:c.1924G>A;p.(Gly642Arg)	Missense	1.17	BM
UPN_45	<i>DNMT3A</i>	NM_175629.2:c.2322+3A>G;p.?	Splice site	1.44	BM
UPN_49	<i>DNMT3A</i>	NM_175629.2:c.2727T>A; p.(Phe909Leu)	Missense	1.85	PB
UPN_49	<i>SF3B1</i>	NM_012433.3:c.1997A>C; p.(Lys666Thr)	Missense	9.01	PB
UPN_5	<i>ASXL1</i>	NM_015338.6:c.1934dup;p.(Gly646Trpfs*12)	InDel	7.01	BM
UPN_50	<i>DNMT3A</i>	NM_175629.2:c.2391C>A;p.(Asn797Lys)	Missense	1.51	BM
UPN_50	<i>NFE2</i>	NM_001136023.3:c.705dup;p.(Pro236Serfs*14)	InDel	3.09	BM
UPN_51	<i>DNMT3A</i>	NM_175629.2:c.2494A>G;p.(Thr832Ala)	Missense	6.42	BM
UPN_51	<i>ZRSR2</i>	NM_005089.3:c.370C>T;p.(Gln124*)	Nonsense	2.93	BM
UPN_52	<i>DNMT3A</i>	NM_175629.2:c.1949T>G, p.(Leu650Arg)	Missense	1.28	BM
UPN_52	<i>TP53</i>	NM_000546.5:c.536A>G, p.(His179Arg)	Missense	4.48	BM
UPN_53	<i>DNMT3A</i>	NM_175629.2: c.2644C>T, p.(Arg882Cys)	Missense	27.42	BM
UPN_53	<i>NFE2</i>	NM_001136023.3:c.578_581del, p.(Asn193Ilefs*12)	InDel	24.62	BM
UPN_54	<i>DNMT3A</i>	NM_175629.2:c.2332G>A;p.(Val778Met)	Missense	1.03	BM
UPN_54	<i>U2AF2</i>	NM_007279.2:c.977G>C;p.(Gly326Ala)	Missense	7.93	BM
UPN_55	<i>DNMT3A</i>	NM_175629.2:c.1668-2A>G;p.?	Splice site	2.32	BM

Subject	Gene	Variant	Mutation type	VAF [%]	Specimen
UPN_55	MYD88	NM_001172567.1:c.818T>C;p.(Leu273Pro)	Missense	27.86	BM
UPN_57	DNMT3A	M_175629.2:c.2096G>T; p.(Gly699Val)	Missense	1.44	BM
UPN_57	DNMT3A	NM_175629.2:c.1911_1914delGTCT;p.(Phe640Metfs*10)	InDel	3.33	BM
UPN_58	DNMT3A	NM_175629.2:c.2656C>T; p.(Gln886*)	Nonsense	1.20	BM
UPN_58	DNMT3A	NM_175629.2:c.1936+1G>T;p.?	Splice site	6.30	BM
UPN_59	DNMT3A	NM_175629.2:c.1071_1093del;p.(Thr358Profs*27)	InDel	5.38	BM
UPN_59	DNMT3A	NM_175629.2:c.2206C>T;p.(Arg736Cys)	Missense	31.70	BM
UPN_60	DNMT3A	NM_175629.2:c.2106_2107insA;p.(Leu703Thrfs*10)	InDel	1.23	BM
UPN_60	DNMT3A	NM_175629.2:c.1501_1508dup;p.(Leu504Metfs*150)	InDel	4.19	BM
UPN_61	DNMT3A	NM_175629.2:c.2045T>G;p.(Met682Arg)	Missense	2.39	BM
UPN_61	DNMT3A	NM_175629.2:c.2245C>T;p.(Arg749Cys)	Missense	3.29	BM
UPN_62	DNMT3A	NM_175629.2:c.2339T>C;p.(Ile780Thr)	Missense	1.22	PB
UPN_62	DNMT3A	NM_175629.2:c.1040T>C;p.(Leu347Pro)	Missense	32.70	PB
UPN_64	DNMT3A	NM_175629.2:c.1154del;p.(Pro385Argfs*22)	InDel	4.88	BM
UPN_64	TP53	NM_000546.5:c.524G>A;p.(Arg175His)	Missense	1.99	BM
UPN_64	U2AF2	NM_007279.2:c.866A>G;p.(Asn289Ser)	Missense	1.58	BM
UPN_65	DNMT3A	NM_175629.2:c.2580G>A;p.(Trp860*)	Nonsense	1.21	PB
UPN_65	DNMT3A	NM_175629.2:c.1851+1G>A;p.?	Splice site	2.57	PB
UPN_65	DNMT3A	NM_175629.2:c.2458G>T;p.(Glu820*)	Nonsense	2.20	PB
UPN_66	DNMT3A	NM_175629.2:c.2644C>T;p.(Arg882Cys)	Missense	1.96	PB
UPN_66	DNMT3A	NM_175629.2:c.1240_1245delTTCCAG;p.(Phe414_Gln415del)	InDel	1.18	PB
UPN_66	IDH2	NM_002168.3:c.419G>A;p.(Arg140Gln)	Missense	7.43	PB
UPN_69	JAK2	NM_004972.3:c.1849G>T p.(Val617Phe)	Missense	9.92	BM
UPN_69	TET2	NM_001127208.2:c.4045-2A>G;p.?	Splice site	3.56	BM
UPN_7	ASXL1	NM_015338.6:c.2644C>T; p.(Gln882*)	Nonsense	1.05	PB
UPN_7	ZBTB7A	NM_015898.4:c.1228T>A;p.(Tyr410Asn)	Missense	2.89	PB
UPN_70	JAK2	NM_004972.3:c.1849G>T p.(Val617Phe)	Missense	3.54	BM
UPN_70	TET2	NM_001127208.2:c.3789T>A;p.(Cys1263*)	Nonsense	2.60	BM
UPN_71	KRAS	NM_033360.3:c.35G>A;p.(Gly12Asp)	Missense	22.19	PB
UPN_71	TET2	NM_001127208.2:c.4393C>T;p.(Arg1465*)	Nonsense	3.02	PB
UPN_72	MYD88	NM_001172567.1:c.818T>C;p.(Leu273Pro)	Missense	2.38	BM
UPN_72	TET2	NM_001127208.2:c.2156del;p.(Leu719Cysfs*32)	InDel	1.21	BM
UPN_72	TET2	NM_001127208.2:c.5629A>G;p.(Lys1877Glu)	Missense	1.78	BM
UPN_72	TET2	NM_001127208.2:c.3647G>A;p.(Arg1216Gln)	Missense	2.42	BM
UPN_73	NOTCH1	ENST00000277541:c.7541_7542delCT;p.Pro2514fs	InDel	1.39	BM
UPN_74	PHF6	NM_032458.3:c.998A>T;p.(Asp333Val)	Missense	1.43	BM
UPN_75	PPM1D	NM_003620.3:c.1545_1546delGT; p.(Met515Ilefs*12)	InDel	1.82	PB
UPN_75	TET2	NM_001127208.2:c.3817T>C; p.(Cys1273Arg)	Missense	3.44	PB
UPN_75	TET2	NM_001127208.2:c.980C>G; p.(Ser327*)	Nonsense	1.50	PB
UPN_76	PPM1D	NM_003620.3:c.1636delC; p.(Leu546*)	InDel	2.66	PB
UPN_77	PPM1D	NM_003620.4:c.1573G>T; p.(Glu525*)	Nonsense	1.38	BM
UPN_78	RUNX1	NM_001754.4:c.664delT; p.(Ser222Profs*15)	InDel	2.69	PB
UPN_78	TET2	NM_001127208.2:c.1081C>T; p.(Gln361*)	Nonsense	1.37	PB

Subject	Gene	Variant	Mutation type	VAF [%]	Specimen
UPN_79	<i>RUNX1</i>	NM_001754.4:c.833C>A;p.(Pro278Gln)	Missense	1.76	BM
UPN_79	<i>TERT</i>	NM_198253.3:c.1573+6G>T;p.?	Splice site	4.96	BM
UPN_8	<i>BCOR</i>	NM_001123385.2:c.4163C>T; p.(Ala1388Val)	Missense	1.23	PB
UPN_8	<i>DNMT3A</i>	NM_175629.2:c.886G>A; p.(Val296Met)	Missense	3.21	PB
UPN_8	<i>TET2</i>	NM_001127208.2:c.4525A>T;p.(Lys1509*)	Nonsense	2.46	PB
UPN_8	<i>ZRSR2</i>	NM_005089.3:c.1338_1343dup;p.(Ser447_Arg448dup)	InDel	31.32	PB
UPN_80	<i>SF3B1</i>	NM_012433.3:c.2230G>C;p.(Ala744Pro)	Missense	2.61	PB
UPN_81	<i>SF3B1</i>	NM_012433.3:c.1873C>T, p.(Arg625Cys)	Missense	14.97	BM
UPN_82	<i>SRSF2</i>	NM_003016.4:c.283C>G;p.(Pro95Ala)	Missense	26.53	BM
UPN_85	<i>TET2</i>	NM_001127208.2:c.3885delC; p.(Tyr1295*)	InDel	2.81	PB
UPN_86	<i>TET2</i>	NM_001127208.2:c.5127dup;p.(Thr1710Tyrfs*3)	InDel	1.66	PB
UPN_87	<i>TET2</i>	NM_001127208.2:c.670G>T;p.(Glu224*)	Nonsense	2.16	PB
UPN_88	<i>TET2</i>	NM_001127208.2:c.456_457dup;p.(Ser153Phefs*3)	InDel	2.24	PB
UPN_89	<i>TET2</i>	NM_001127208.2:c.3954+5G>A;p.?	Splice site	2.10	BM
UPN_9	<i>BCOR</i>	NM_001123385.2:c.838G>A;p.(Val280Ile)	Missense	13.32	BM
UPN_9	<i>CXCR4</i>	NM_001008540.2:c.1045dup;p.(Glu349Glyfs*13)	InDel	1.92	BM
UPN_90	<i>TET2</i>	NM_001127208.2:c.4353del;p.(Arg1452Glufs*6)	InDel	5.17	BM
UPN_91	<i>TET2</i>	NM_001127208.2:c.5079_5082del;p.(Tyr1693*)	InDel	7.13	BM
UPN_92	<i>TET2</i>	NM_001127208.2:c.1486del;p.(Met496*)	InDel	1.30	BM
UPN_96	<i>TET2</i>	NM_001127208.2:c.3662G>A;p.(Cys1221Tyr)	Missense	3.24	PB
UPN_96	<i>TET2</i>	NM_001127208.2:c.3482G>C;p.(Arg1161Thr)	Missense	5.00	PB
UPN_97	<i>TET2</i>	NM_001127208.2:c.5650A>G;p.(Thr1884Ala)	Missense	1.11	BM
UPN_97	<i>TET2</i>	NM_001127208.2:c.4161C>A;p.(Asn1387Lys)	Missense	15.24	BM
UPN_98	<i>TET2</i>	NM_001127208.2:c.663_666delACAT;p.(His222Valfs*27)	InDel	2.14	PB
UPN_98	<i>TET2</i>	NM_001127208.2:c.1038_1039delAG;p.(Ala347Valfs*3)	InDel	3.27	PB
UPN_99	<i>U2AF2</i>	NM_007279.2:c.259_261delAAG;p.(Lys87del)	InDel	1.08	BM

Abbreviations: VAF, variant allele frequency; InDel, insertion/deletion variant; BM, bone marrow; PB, peripheral blood; UPN, unique patient number.

Supplemental Table 3: Comparison of variant allele frequencies in paired BM and PB samples from 21 individuals with clonal hematopoiesis.

Subject	Gene	Variant	BM sample		PB sample	
			VAF [%]	Coverage	VAF [%]	Coverage
UPN_6	ASXL1	c.2512_2537dup; p.(Ser846Argfs*5)	16.92	1876	11.42	1655
	ETV6	c.844dup;p.(Arg282Profs*18)	0.37	2685	0.58	1719
UPN_7	ASXL1	c.2644C>T; p.(Gln882*)	6.95	820	1.05	1045
	ZBTB7A	c.1228T>A;p.(Tyr410Asn)	2.50	1482	2.89	1383
UPN_8	BCOR*	c.4163C>T; p.(Ala1388Val)	ND	700	1.22	734
	TET2	c.4525A>T;p.(Lys1509*)	7.06	1126	2.46	1342
	DNMT3A	c.886G>A; p.(Val296Met)	1.62	618	3.21	654
	DNMT3A	c.2339T>G;p.(Ile780Ser)	1.51	1323	0.06	3451
	ZRSR2	c.1338_1343dup;p.(Ser447_Arg448dup)	43.06	353	31.32	514
UPN_18	DNMT3A	c.2092T>G; p.(Trp698Gly)	6.97	717	5.77	762
UPN_19	DNMT3A	c.2195T>C; p.(Phe732Ser)	1.32	683	1.15	786
UPN_21	DNMT3A	c.2047_2056del; p.(Tyr683Thrfs*19)	4.55	857	3.41	586
UPN_29	DNMT3A	c.2322+2T>C;p.?	0.64	1713	1.35	1111
UPN_31	DNMT3A	c.1904G>A;p.(Arg635Gln)	6.33	1248	6.54	1469
UPN_49	DNMT3A	c.2727T>A; p.(Phe909Leu)	2.37	1054	1.85	758
	SF3B1	c.1997A>C; p.(Lys666Thr)	9.88	921	9.01	677
UPN_65	DNMT3A	c.2458G>T; p.(Glu820*)	3.18	1634	2.20	1407
	DNMT3A	c.1851+1G>A; p.?	2.17	738	2.57	818
	DNMT3A	c.2580G>A;p.(Trp860*)	1.29	851	1.21	745
UPN_66	DNMT3A	c.1240_1245delTTCCAG;p.(Phe414_Gln415del)	1.91	1467	1.13	704
	DNMT3A	c.2644C>T;p.(Arg882Cys)	3.91	818	1.96	1124
	IDH2	c.419G>A;p.(Arg140Gln)	2.67	1422	7.43	1790
	DNMT3A	c.1429+1G>A;p.?	1.96	1018	0.66	1958
UPN_71	TET2	c.4393C>T;p.(Arg1465*)	3.07	489	3.02	364
	KRAS	c.35G>A;p.(Gly12Asp)	16.50	721	22.19	1005
UPN_75	TET2	c.980C>G; p.(Ser327*)	1.79	1119	1.50	1203
	TET2	c.3817T>C; p.(Cys1273Arg)	4.22	1207	3.44	1017
	PPM1D	c.1545_1546delGT; p.(Met515Ilefs*12)	3.75	773	1.82	881
UPN_76	PPM1D	c.1636delC; p.(Leu546*)	3.11	644	2.66	826
UPN_80	SF3B1	c.2230G>C;p.(Ala744Pro)	6.35	929	2.61	881
UPN_86	TET2	c.5127dup;p.(Thr1710Tyrfs*3)	1.66	1743	1.66	2046
UPN_88	TET2	c.456_457dup;p.(Ser153Phefs*3)	4.19	1337	2.24	848
UPN_96	TET2	c.3662G>A;p.(Cys1221Tyr)	3.29	3682	3.24	2408
	TET2	c.3482G>C;p.(Arg1161Thr)	9.84	2257	5.00	1341
	DNMT3A	c.1945G>T;p.(Val649Leu)	1.42	1615	0.73	2042
	TET2	c.822del;p.(Asn275Ilefs*18)	2.63	3226	0.67	4480
UPN_98	DNMT3A*	c.731del;p.(Pro244Leufs*72)	1.21	828	ND	874
	TET2	c.663_666delACAT;p.(His222Valfs*27)	1.40	714	2.14	1167
	TET2	c.1038_1039delAG;p.(Ala347Valfs*3)	2.23	1165	3.27	1588
UPN_27 2	SRP72	c.869A>G;p.(Asn290Ser)	4.07	589	3.16	792
	TET2	c.3883T>G;p.(Tyr1295Asp)	1.28	1249	0.75	2259
UPN_27 6	DNMT3A	c.1238dup;p.(Phe414Leufs*7)	1.86	1557	1.42	1973
	STAG2	p.(Leu203Phe)	1.47	955	0.34	2336

* Highlighted variants were validated by digital droplet PCR, results are shown in Supplemental Table 4

Abbreviations: VAF, variant allele frequency; InDel, insertion/deletion variant; BM, bone marrow, PB, peripheral blood; ND, not detectable; UPN, unique patient number.

Supplemental Table 4: Orthogonal validation by mutation-specific digital droplet PCR of variants identified in paired BM and PB samples (UPN_98 and UPN_8) and in paired bone marrow samples from individuals undergoing simultaneous bilateral hip replacement (UPN_2 and UPN_248).

Subject	Gene	Variant	Sample	VAF [%]
UPN_98	<i>DNMT3A</i>	c.731del; p.(Pro244Leufs*72)	BM	0.831
			PB	0.084
UPN_8	<i>DNMT3A</i>	c.2339T>G; p.(Ile780Ser)	BM	1.584
			PB	0.267
UPN_2	<i>ASXL1</i>	c.2083C>T; p.(Gln695*)	Hip 1	1.087
			Hip 2	0.050
UPN_248	<i>ASXL1</i>	c.2258_2259dup; p.(Thr754Profs*19)	Hip 1	0.274
			Hip 2	4.154

Abbreviations: VAF, variant allele frequency; BM, bone marrow, PB, peripheral blood; UPN, unique patient number.

Supplemental Table 5: Overview of variants found in patients with ‘large-clone’ CHIP.

Large-clone CHIP was defined as at least one variant with a VAF >20% (highlighted in red in the table). Subjects are grouped according to their clonal profile (single variants, multiple variants with one “dominant” variant, or several variants with VAF >20%).

Mutational profile	UPN	Gene	Variant	Mutation type	VAF [%]	
Single variant	UPN_82	<i>SRSF2</i>	NM_003016.4:c.283C>G;p.(Pro95Ala)	Missense	26.5	
	UPN_20	<i>DNMT3A</i>	NM_175629.2:c.2591T>A; p.(Met864Lys)	Missense	21.3	
Multiple variants	UPN_254	<i>TET2</i>	NM_001127208.2:c.5650A>G;p.(Thr1884Ala)	Missense	43.9	
		<i>SRSF2</i>	NM_003016.4:c.242A>T;p.(Asp81Val)	Missense	3.2	
	UPN_62	<i>DNMT3A</i>	NM_175629.2:c.1040T>C;p.(Leu347Pro)	Missense	32.7	
		<i>DNMT3A</i>	NM_175629.2:c.2339T>C;p.(Ile780Thr)	Missense	1.2	
	UPN_59	<i>DNMT3A</i>	NM_175629.2:c.2206C>T;p.(Arg736Cys)	Missense	31.7	
		<i>DNMT3A</i>	NM_175629.2:c.1071_1093del;p.(Thr358Profs*27)	InDel	5.4	
	UPN_8	<i>ZRSR2</i>	NM_005089.3:c.1338_1343dup;p.(Ser447_Arg448dup)	InDel	31.2	
		<i>DNMT3A</i>	NM_175629.2:c.886G>A; p.(Val296Met)	Missense	3.2	
		<i>TET2</i>	NM_001127208.2:c.4525A>T;p.(Lys1509*)	Nonsense	2.5	
		<i>BCOR</i>	NM_001123385.2:c.4163C>T; p.(Ala1388Val)	Missense	1.2	
	UPN_55	<i>MYD88</i>	NM_001172567.1:c.818T>C;p.(Leu273Pro)	Missense	27.9	
		<i>DNMT3A</i>	NM_175629.2:c.1668-2A>G;p.?	Splice site	2.3	
	UPN_15	<i>DNMT3A</i>	NM_175629.2:c.1123-2A>G;p.?	Splice site	23.6	
		<i>TET2</i>	NM_001127208.2:c.4079T>G;p.(Leu1360Arg)	Missense	1.2	
	UPN_71	<i>KRAS</i>	NM_033360.3:c.35G>A;p.(Gly12Asp)	Missense	22.3	
		<i>TET2</i>	NM_001127208.2:c.4393C>T;p.(Arg1465*)	Nonsense	3.0	
	multiple variants with high allelic burden	UPN_248	<i>SH2B3</i>	NM_005475.2:c.1522dup;p.(Arg508Profs*38)	InDel	40.4
			<i>TET2</i>	NM_001127208.2:c.5481_5482del;p.(Gln1828Alafs*17)	InDel	32.3
<i>TET2</i>			NM_001127208.2:c.5165del;p.(Pro1722Leufs*23)	InDel	30.1	
<i>ASXL1</i>			NM_015338.6:c.2258_2259dup;p.(Thr754Profs*19)	InDel	4.9	
UPN_53		<i>DNMT3A</i>	NM_175629.2: c.2644C>T, p.(Arg882Cys)	Missense	27.4	
		<i>NFE2</i>	NM_001136023.3:c.578_581del, p.(Asn193Ilefs*12)	InDel	24.6	

Abbreviations: VAF, variant allele frequency; InDel, insertion/deletion variant; UPN, unique patient number.

Supplemental Table 6: Paired-sample sequencing results for 11 patients undergoing bilateral hip replacements.

Hip pair	Variant	Hip 1		Hip 2		<i>p</i> (Fisher's exact test)
		Variant reads	VAF [%]	Variant reads	VAF [%]	
UPN_2	ASXL1:c.2083C>T;p.(Gln695*)	29/2307	1.29	2/4015	NA	<.0001
	JAK2:c.1849G>T, p.(Val617Phe)	94/1577	5.96	141/2589	5.45	.49
	DNMT3A:c.2141C>G;p.(Ser714Cys)	20/2358	0.85	67/5779	1.15	.24
UPN_248	ASXL1:c.2258_2259dup;p.(Thr754Profs*19)	9/2779	0.32	144/2950	4.88	<.0001
	TET2:c.5165del;p.(Pro1722Leufs*23)	432/1485	29.09	450/1495	30.1	.55
	TET2:c.5481_5482del;p.(Gln1828Alafs*17)	549/1644	33.39	575/1780	32.3	.51
	SH2B3:c.1522dup;p.(Arg508Profs*38)	565/1535	36.81	679/1680	40.42	.04
UPN_242	ASXL1:c.2885_2886del;p.(Val962Alafs*7)	18/1402	1.28	40/2143	1.87	.22
UPN_243	TET2:c.2919C>A;p.(Cys973*)	26/1544	1.68	9/477	1.89	.84
UPN_73	NOTCH1:c.7541_7542delCT;p.Pro2514fs	11/552	1.99	7/503	1.39	.49
UPN_26	DNMT3A:c.2332G>A;p.(Val778Met)	76/1498	5.07	80/1688	4.74	.68
UPN_251	TET2:c.5618T>C;p.(Ile1873Thr)	25/2851	0.88	28/2252	1.15	.21
	DNMT3A:c.2530A>G;p.(Lys844Glu)	13/1447	0.90	20/1285	1.45	.16
UPN_250	TET2:c.2375C>A;p.(Ser792*)	23/1988	1.1	17/1785	0.95	.63
UPN_249	No variants identified					NA
UPN_240	No variants identified					NA
UPN_241	No variants identified					NA

Footnotes: Variants that were detected by manual inspection of sequencing reads are highlighted in red.

Abbreviations: VAF, variant allele frequency; NA, not applicable; UPN, unique patient number.

Supplemental Table 7: Allelic burden of eight flow-sorted subpopulations and bulk of five *ASXL1* mutated individuals.

Subject	<i>ASXL1</i> (NM_015338.6) variant	VAF in flow-sorted cell fractions								
		bulk	CD34 ⁺	HSC	CMP	MEP	GMP	CD15 ⁺	CD3 ⁺	CD19 ⁺
UPN_226	c.2179G>T;p.(Glu727*)	1.70	2.94	2.33	3.97	6.00	2.29	1.42	0.00	0.53
UPN_1	c.2036dup;p.(Gly680Argfs*38)	3.31	3.81	2.64	5.56	9.86	3.79	7.91	0.23	0.54
	c.2385del;p.(Trp796Glyfs*22)	4.72	8.93	8.79	11.42	9.68	5.48	7.57	0.00	0.00
UPN_4	c.1934dup;p.(Gly646Trpfs*12)	7.38	11.60	12.23	12.70	19.67	10.60	7.58	5.12	11.11
UPN_5	c.1934dup;p.(Gly646Trpfs*12)	9.24	12.18	13.37	12.93	5.13	10.64	4.83	1.23	8.82
UPN_3	c.2954_2957del;p.(Ile985Thrfs*7)	4.90	6.15	18.11	13.58	6.48	2.56	1.17	0.00	0.34

Abbreviations: HSC, hematopoietic stem cells; CMP, common-myeloid progenitors; MEP, megakaryocyte-erythroid progenitors; GMP, granulocyte-monocyte progenitors; VAF, variant allele frequency; UPN, unique patient number.

Supplemental Table 8: Statistical analyses of differential clonal involvement of flow-sorted subpopulations and bulk marrow in *ASXL1*-mutated individuals.

Corrected *p*-values are indicated for the analysis of each pair of subpopulations using Fisher's test followed by correction for multiple hypothesis testing using the Benjamini-Hochberg procedure. Significant differences are indicated in red.

		HSC	CD34+	CMP	GMP	MEP	CD3+	CD19+	CD15+	bulk
UPN_226	NM_015338.6:c.2179G>T; p.(Glu727*)	HSC								
		CD34+	1.00							
		CMP	0.81	0.67						
		GMP	1.00	0.74	0.49					
		MEP	0.44	0.26	0.55	0.21				
		CD3+	0.052	0.00011	6.83x10 ⁻⁵	0.0031	0.00011			
		CD19+	0.25	0.016	0.0032	0.099	0.0032	0.21		
		CD15+	0.74	0.27	0.10	0.55	0.052	0.017	0.29	
		bulk	0.74	0.27	0.099	0.73	0.052	0.0031	0.16	0.86
		UPN_1	NM_015338.6:c.2036dup; p.(Gly680Argfs*38)	HSC						
CD34+	0.61									
CMP	0.15			0.15						
GMP	0.61			1.00	0.22					
MEP	0.0046			0.0016	0.057	0.0045				
CD3+	0.0012			3.24x10 ⁻¹⁰	6.21x10 ⁻¹⁴	1.59x10 ⁻⁸	8.70x10 ⁻¹⁵			
CD19+	0.017			2.46x10 ⁻⁶	3.17x10 ⁻⁹	3.29x10 ⁻³	4.07x10 ⁻¹¹	0.36		
CD15+	0.011			0.0058	0.18	0.013	0.55	7.07x10 ⁻¹⁷	3.77x10 ⁻¹²	
bulk	0.86			0.68	0.051	0.71	0.0039	3.17x10 ⁻⁹	2.16x10 ⁻⁵	0.00084
UPN_4	NM_015338.6:c.1934dup; p.(Gly646Trpfs*12)			HSC						
		CD34+	0.92							
		CMP	0.97	0.78						
		GMP	0.64	0.78	0.46					
		MEP	0.016	0.0058	0.0058	0.0011				
		CD3+	0.0054	0.0058	0.00041	0.016	1.16x10 ⁻⁹			
		CD19+	0.90	1.00	0.78	0.97	0.021	0.037		
		CD15+	0.15	0.19	0.065	0.35	5.57x10 ⁻⁵	0.35	0.35	
		bulk	0.063	0.072	0.011	0.19	7.08x10 ⁻⁸	0.29	0.22	1.00
		UPN_5	NM_015338.6:c.1934dup; p.(Gly646Trpfs*12)	HSC						
CD34+	0.74									
CMP	0.91			0.80						
GMP	0.67			0.75	0.71					
MEP	0.0097			0.0046	0.0026	0.097				
CD3+	1.56x10 ⁻¹⁰			1.15x10 ⁻¹⁵	1.59x10 ⁻¹⁶	2.5x10 ⁻⁶	0.0044			
CD19+	0.097			0.088	0.038	0.67	0.13	2.86x10 ⁻¹⁰		
CD15+	0.0020			0.00041	9.00x10 ⁻⁵	0.050	0.87	0.0026	0.041	
bulk	0.15			0.13	0.077	0.74	0.088	1.75x10 ⁻¹⁰	0.87	0.024
UPN_3	NM_015338.6:c.2954_2957del; p.(Ile985Thrfs*7)			HSC						
		CD34+	2.64x10 ⁻⁷							
		CMP	0.054	0.0016						
		GMP	3.45x10 ⁻¹⁵	0.031	9.09x10 ⁻⁹					
		MEP	1.82x10 ⁻⁵	0.87	0.0039	0.027				
		CD3+	1.11x10 ⁻²⁷	7.83x10 ⁻⁸	1.29x10 ⁻¹⁸	0.0010	6.13x10 ⁻⁶			
		CD19+	1.77x10 ⁻³¹	1.79x10 ⁻⁷	3.29x10 ⁻²⁰	0.0068	1.41x10 ⁻⁷	0.53		
		CD15+	2.04x10 ⁻¹⁴	0.0034	2.85x10 ⁻⁹	0.30	0.0028	0.061	0.20	
		bulk	2.16x10 ⁻¹⁹	0.42	2.32x10 ⁻⁸	0.060	0.33	3.14x10 ⁻⁸	3.98x10 ⁻⁸	0.0058
		UPN_1	NM_015338.6:c.2385del; p.(Trp796Glyfs*22)	HSC						
CD34+	1.00									
CMP	0.74			0.57						
GMP	0.49			0.31	0.048					
MEP	1.00			0.90	0.90	0.38				
CD3+	2.00x10 ⁻⁶			1.29x10 ⁻¹⁰	1.32x10 ⁻¹²	4.34x10 ⁻⁵	8.39x10 ⁻⁶			
CD19+	9.69x10 ⁻⁶			8.04x10 ⁻⁰⁹	1.24x10 ⁻¹⁰	2.72x10 ⁻⁵	2.72x10 ⁻⁵	1.00		
CD15+	0.90			0.88	0.32	0.58	0.77	1.37x10 ⁻⁷	1.60x10 ⁻⁵	
bulk	0.24			0.048	0.0030	0.88	0.24	1.02x10 ⁻⁶	1.55x10 ⁻⁵	0.32

Abbreviations: HSC, hematopoietic stem cells; CMP, common-myeloid progenitors; MEP, megakaryocyte-erythroid progenitors; GMP, granulocyte-monocyte progenitors; VAF, variant allele frequency; UPN, unique patient number.

Supplemental Table 9: Longitudinal analysis in the baseline non-CH cohort over a time of up to 24 months.

Variants highlighted in red were not called during primary data analysis, but detected upon manual inspection of sequencing reads.

Subject	Gene	Variant	VAF [%]				
			Screening	6 mo	12 mo	18 mo	24 mo
UPN_281		none	none	none	NA	NA	NA
UPN_284		none	none	none	none	NA	NA
UPN_285		none	none	none	none	NA	NA
UPN_102	<i>DNMT3A</i>	NM_175629.2:c.[1257delT]; p.(Lys420Argfs*231)	0.70	NA	1.32	NA	2.00
UPN_103		none	none	NA	none	none	NA
UPN_104		none	none	NA	none	NA	NA
UPN_105		none	none	NA	none	NA	NA
UPN_106	<i>PRPF8</i>	NM_006445.4:c.3361A>G; p.(Asn1121Asp)	0.91	0.82	1.02	1.20	NA
UPN_107		none	none	NA	none	NA	NA
UPN_108		none	none	NA	none	NA	NA
UPN_109		none	none	NA	none	NA	NA
UPN_110		none	none	none	NA	NA	NA
UPN_111		none	none	none	NA	NA	NA
UPN_112		none	none	NA	none	NA	NA
UPN_113		none	none	none	none	NA	NA
UPN_114		none	none	NA	none	NA	NA
UPN_115		none	none	none	NA	NA	NA
UPN_116		none	none	none	none	NA	NA
UPN_117		none	none	none	none	NA	NA
UPN_118		none	none	none	NA	NA	NA
UPN_119		none	none	none	NA	NA	NA
UPN_120	<i>ASXL1</i>	NM_015338.6:c.2036dup; p.(Gly680Argfs*38)	1.16	1.78	NA	NA	NA
	<i>TET2</i>	NM_001127208.2:c.3986T>C; p.(Leu1329Pro)	1.05	1.42	NA	NA	NA
UPN_121		none	none	none	NA	NA	NA
UPN_122		none	none	none	none	NA	NA
UPN_124		none	none	NA	none	NA	NA
UPN_126		none	none	none	NA	NA	NA
UPN_127		none	none	NA	none	NA	NA

Abbreviations: VAF, variant allele frequency; mo, months; NA, not analyzed; UPN, unique patient number.

Supplemental Table 10: Longitudinal analysis of CH temporal evolution over a time of up to 18 months.

Bold font indicates variants that emerged or dropped below the limit of detection during follow-up. Variants in red were not called during primary data analysis, but detected upon manual inspection of sequencing reads.

Subject	Gene	Variant	VAF [%]			
			Screening	6 mo	12 mo	18 mo
UPN_49	<i>DNMT3A</i>	c.2727T>A; p.(Phe909Leu)	1.85	1.19	2.20	NA
	<i>SF3B1</i>	c.1997A>C; p.(Lys666Thr)	9.01	6.97	9.25	NA
UPN_85	<i>TET2</i>	c.3885delC; p.(Tyr1295*)	2.81	3.71	5.05	4.53
UPN_18	<i>DNMT3A</i>	c.2092T>G; p.(Trp698Gly)	5.77	7.57	4.75	4.54
UPN_75	<i>TET2</i>	c.980C>G; p.(Ser327*)	1.50	2.42	1.41	1.85
	<i>TET2</i>	c.3817T>C; p.(Cys1273Arg)	3.44	3.05	4.33	3.95
	<i>PPM1D</i>	c.1545_1546delGT; p.(Met515Ilefs*12)	1.82	2.93	3.56	2.9
UPN_65	<i>DNMT3A</i>	c.2458G>T; p.(Glu820*)	2.20	1.70	2.99	NA
	<i>DNMT3A</i>	c.1851+1G>A;p.?	2.57	3.70	3.99	NA
	<i>DNMT3A</i>	c.2580G>A;p.(Trp860*)	1.21	1.00	0.39	NA
UPN_6	<i>ASXL1</i>	c.2512_2537dup; p.(Ser846Argfs*5)	11.42	11.320	9.19	NA
	<i>ETV6</i>	c.844dup;p.(Arg282Profs*18)	0.58	1.40	0.30	NA
UPN_19	<i>DNMT3A</i>	c.2195T>C; p.(Phe732Ser)	1.15	0.89	0.95	NA
UPN_20	<i>DNMT3A</i>	c.2591T>A; p.(Met864Lys)	21.29	15.28	9.38	NA
UPN_21	<i>DNMT3A</i>	c.2047_2056del; p.(Tyr683Thrfs*19)	3.41	5.23	3.68	NA
UPN_76	<i>PPM1D</i>	c.1636delC; p.(Leu546*)	2.66	1.31	2.04	NA
UPN_23	<i>DNMT3A</i>	c.1506delT; p.(Thr503Profs*148)	4.73	4.07	NA	NA
UPN_80	<i>SF3B1</i>	c.2230G>C;p.(Ala744Pro)	2.61	8.73	3.80	NA
UPN_78	<i>TET2</i>	c.1081C>T; p.(Gln361*)	1.37	NA	1.20	NA
UPN_32	<i>DNMT3A</i>	c.2204A>G;p.(Tyr735Cys)	2.60	1.04	2.64	NA
UPN_12	<i>DNMT3A</i>	c.2567_2568delAG;p.(Glu856Glyfs*7)	1.56	0.56	NA	NA
UPN_87	<i>TET2</i>	c.670G>T;p.(Glu224*)	2.16	1.92	NA	NA
UPN_86	<i>TET2</i>	c.5127dup;p.(Thr1710Tyrfs*3)	1.66	1.89	NA	NA
UPN_29	<i>DNMT3A</i>	c.2322+2T>C;p.?	1.35	NA	NA	1.13
UPN_282	<i>DNMT3A</i>	c.2190_2215del;p.(Phe731*)	10.81	20.92	NA	NA
UPN_283	<i>DNMT3A</i>	c.890G>C;p.(Trp297Ser)	1.65	2.31	NA	NA
	<i>DNMT3A</i>	c.2207G>A;p.(Arg736His)	5.68	6.87	NA	NA
	<i>SF3B1</i>	c.1997A>G;p.(Lys666Arg)	2.69	3.70	NA	NA
	<i>ASXL1</i>	c.1934dup p.(Gly646Trpfs*12)	0.68	1.16	NA	NA
UPN_7	<i>ASXL1</i>	c.2644C>T; p.(Gln882*)	1.05	NA	ND	NA
	<i>ZBTB7A</i>	c.1228T>A;p.(Tyr410Asn)	2.89	NA	1.88	NA

Abbreviations: VAF, variant allele frequency; ND, not detected; mo, months; NA, not analyzed; UPN, unique patient number

Supplemental Table 11: Variants identified in patients with MDS or sAML.

Subject	Gene	Variant	Mutation type	VAF [%]
MDS_1	<i>CBL</i>	NM_005188.3:c.1244G>A;p.(Gly415Asp)	Missense	38.0
MDS_1	<i>TET2</i>	NM_001127208.2:c.3404G>A;p.(Cys1135Tyr)	Missense	36.8
MDS_1	<i>TP53</i>	NM_000546.5:c.827C>G;p.(Ala276Gly)	Missense	25.1
MDS_2	<i>DDX41</i>	NM_016222:c.1574G>A;p.R525H	Missense	4.3
MDS_2	<i>DNMT3A</i>	NM_175629.2: c.2645G>A p.(Arg882His)	Missense	5.1
MDS_2	<i>SRSF2</i>	NM_003016.4:c.284C>A p.(Pro95His)	Missense	4.7
MDS_3	<i>BCOR</i>	NM_001123385.2:c.3649del;p.(Arg1217Aspfs*21)	InDel	32.3
MDS_3	<i>BCORL1</i>	NM_021946.4:c.4230dup;p.(Glu1411*)	InDel	4.9
MDS_3	<i>DNMT3A</i>	NM_175629.2:c.2644C>A;p.(Arg882Ser)	Missense	19.3
MDS_3	<i>NRAS</i>	NM_002524.5:c.38G>T;p.(Gly13Val)	Missense	14.3
MDS_3	<i>U2AF1</i>	NM_001025203.1:c.101C>T;p.(Ser34Phe)	Missense	20.3
MDS_4	<i>RAD21</i>	NM_006265.3:c.742C>G;p.(Leu248Val)	Missense	37.1
MDS_4	<i>SF3B1</i>	NM_012433.3:c.2098A>G p.(Lys700Glu)	Missense	42.6
MDS_4	<i>TET2</i>	NM_001127208.2:c.3854_3856del;p.(Phe1285del)	InDel	30.5
MDS_4	<i>TET2</i>	NM_001127208.2:c.3197_3198dup;p.(Arg1067Leufs*16)	InDel	34.0
MDS_4	<i>TP53</i>	NM_000546.5:c.994-3C>G;p.?	Splice site	1.1
MDS_5	<i>BCOR</i>	NM_001123385.2:c.2340dup;p.(Thr781Hisfs*36)	InDel	9.5
MDS_5	<i>ETNK1</i>	NM_018638.4:c.731A>G;p.(Asn244Ser)	Missense	5.5
MDS_5	<i>RUNX1</i>	NM_001754.4:c.431T>C;p.(Leu144Pro)	Missense	18.8
MDS_5	<i>SRSF2</i>	NM_003016.4:c.284C>G p.(Pro95Arg)	Missense	25.8
MDS_5	<i>TET2</i>	NM_001127208.2:c.3908G>A;p.(Ser1303Asn)	Missense	16.1
MDS_5	<i>TET2</i>	NM_001127208.2:c.1630C>T;p.(Arg544*)	Nonsense	27.0
MDS_6	<i>IDH1</i>	NM_005896.3: c.394C>T p.(Arg132Cys)	Missense	4.3
MDS_7	<i>SRSF2</i>	NM_003016.4:c.284C>A p.(Pro95His)	Missense	11.6
MDS_7	<i>TET2</i>	NM_001127208.2:c.2147C>G;p.(Ser716*)	Nonsense	10.3
MDS_7	<i>RUNX1</i>	NM_001754.4:602G>A p.(Glu138Lys)	Missense	11.9
MDS_7	<i>STAG2</i>	NM_001042750.2:c.1644dup;p.(Thr549Tyrfs*11)	InDel	20.6
MDS_7	<i>BCOR</i>	NM_001123385.2:c.2972_2976del;p.(Leu991Profs*25)	InDel	21.9
MDS_8	<i>BCORL1</i>	NM_021946.4:c.1202C>T;p.(Thr401Met)	Missense	19.5
MDS_8	<i>GATA2</i>	NM_001145661.2:c.952G>A;p.(Ala318Thr)	Missense	17.3
MDS_8	<i>RUNX1</i>	NM_001754.4:c.749delinsCC;p.(Arg250Profs*11)	InDel	15.7
MDS_8	<i>SF3B1</i>	NM_012433.3:c.2098A>G p.(Lys700Glu)	Missense	18.0
MDS_9	<i>SF3B1</i>	NM_012433.3:c.2098A>G p.(Lys700Glu)	Missense	52.5
MDS_9	<i>TET2</i>	NM_001127208.2:c.4636C>T;p.(Gln1546*)	Nonsense	43.7
MDS_10	<i>TET2</i>	NM_001127208.2:c.4210C>T;p.(Arg1404*)	Nonsense	2.9
MDS_10	<i>TET2</i>	NM_001127208.2:c.3781C>T;p.R1261C	Missense	20.3
MDS_10	<i>ZRSR2</i>	NM_005089.3:c.900_901del;p.(Glu300Aspfs*16)	InDel	39.0
MDS_11	<i>ASXL1</i>	NM_015338.6:c.2904dup;p.(Asp969*)	InDel	20.6
MDS_11	<i>TET2</i>	NM_001127208.2:c.4183-2A>G;p.?	Splice site	2.4
MDS_11	<i>TET2</i>	NM_001127208.2:c.3532del;p.E1178Kfs*48	InDel	17.6
MDS_11	<i>TET2</i>	NM_001127208.2:c.4546C>T;p.(Arg1516*)	Nonsense	22.4
MDS_11	<i>TP53</i>	NM_000546.5:c.665C>T;p.(Pro222Leu)	Missense	40.2
MDS_11	<i>U2AF1</i>	NM_001025203.1:c.470A>C p.(Gln157Pro)	Missense	20.2

Subject	Gene	Variant	Mutation type	VAF [%]
MDS_12	ASXL1	NM_015338.6:c.1900_1922del;p.(Glu635Argfs*15)	InDel	2.1
MDS_12	ASXL1	NM_015338.6:c.2082_2092del;p.(Gln695Alafs*19)	InDel	15.3
MDS_12	ETNK1	NM_018638.4:c.731A>G;p.(Asn244Ser)	Missense	5.3
MDS_12	U2AF1	NM_001025203.1:c.470A>C p.(Gln157Pro)	Missense	20.2
MDS_13	SETBP1	NM_015559.3:c.2602G>A;p.Asp868Asn	Missense	4.7
MDS_13	SETBP1	NM_015559.3:c.2608G>A (p.Gly870Ser)	Missense	21.9
MDS_13	SF3B1	NM_012433.3:c.2098A>G p.(Lys700Glu)	Missense	26.4
MDS_13	TET2	NM_001127208.2:c.4956del;p.(Gln1652Hisfs*43)	InDel	1.1
MDS_13	TET2	NM_001127208.2:c.4097G>A;R1366H	Missense	1.8
MDS_13	TET2	NM_001127208.2:c.5071del;p.(Ser1691Leufs*4)	InDel	7.7
MDS_14	SRSF2	NM_003016.4:c.284C>T p.(Pro95Leu)	Missense	13.8
MDS_14	TET2	NM_001127208.2:c.1776T>G;p.(Tyr592*)	Nonsense	22.6
MDS_14	TET2	NM_001127208.2:c.2193del;p.(Gln731Hisfs*20)	InDel	24.8
MDS_14	RUNX1	NM_001754.4:c.1274C>T;p.(Pro425Leu)	Missense	14.2
MDS_15	TET2	NM_001127208.2:c.4108G>A;p.(Gly1370Arg)	Missense	3.8
MDS_15	DNMT3A	NM_175629.2:c.1131del;p.(Ser377Argfs*30)	InDel	1.6
MDS_15	DNMT3A	NM_175629.2:c.1628G>C;p.(Gly543Ala)	Missense	6.0
MDS_15	TP53	NM_000546.5:c.377A>G;p.(Tyr126Cys)	Missense	3.7
MDS_15	KRAS	NM_033360.4:c.35G>C (p.Gly12Ala)	Missense	1.1
MDS_16	SRSF2	NM_003016.4:c.284C>T p.(Pro95Leu)	Missense	17.2
MDS_16	RUNX1	NM_001754.4:c.422C>T;p.(Ser141Leu)	Missense	1.9
MDS_16	RUNX1	NM_001754.4:c.412G>T;p.(Glu138*)	Nonsense	23.6
MDS_17	MPL	NM_005373.2:c.1771T>G;p.(Tyr591Asp)	Missense	41.0
MDS_18	DNMT3A	NM_175629.2:c.2645G>C; p.(Arg882Pro)	Missense	28.0
MDS_18	TET2	NM_001127208.2:c.2553del; p.(Glu852Asnfs*21)	InDel	30.0
MDS_18	TET2	NM_001127208.2:c.2243del; p.(Leu748Tyrfs*3)	InDel	20.0
MDS_18	TET2	NM_001127208.2:c.3784C>T; p.(Arg1262Trp)	Missense	7.6
MDS_18	SRSF2	NM_003016.4:c.284C>T p.(Pro95Leu)	Missense	1.5
MDS_19	ASXL1	NM_015338.5:c.1900_1922del; p.(Glu635Argfs*15)	InDel	35.0
MDS_19	EZH2	NM_004456.4:c.1981A>T; p.(Lys661*)	Nonsense	45.0
MDS_19	RUNX1	NM_001754.4:c.492_493insGG; p.(Arg166Valfs*11)	InDel	13.0
MDS_19	STAG2	NM_001042749.2:c.1117-1G>C;p.?	Splice site	9.0
MDS_19	TET2	NM_001127208.2:c.4075C>T; p.(Arg1359Cys)	Missense	12.0
MDS_19	TET2	NM_001127208.2:c.1326delC; p.(Asn442Lysfs*5)	InDel	2.0
MDS_20	ASXL1	NM_015338.5:c.1934dup (p.Gly646Trpfs*12)	InDel	22.0
MDS_20	BCOR	NM_001123385.1:c.359C>G; p.(Pro120Arg)	Missense	52.0
MDS_20	JAK2	NM_004972.3:c.1972G>T; p.(Ala658Ser)	Missense	37.0
MDS_20	RUNX1	NM_001754.4:c.1167dup; p.(Gln390Alafs*210)	InDel	34.0
MDS_21	ASXL1	NM_015338.5:c.1900_1922del; p.(Glu635Argfs*15)	InDel	22.0
MDS_21	CBL	NM_005188.3:c.1249C>A; p.(Pro417Thr)	Missense	21.0
MDS_21	ETNK1	NM_018638.4:c.728A>T; p.(His243Leu)	Missense	18.0
MDS_21	EZH2	NM_004456.4:c.540delA; p.(Gln180Hisfs*61)	InDel	36.0
MDS_21	TET2	NM_001127208.2:c.3594+3A>C;p.?	Splice site	20.0
MDS_22	SRSF2	NM_003016.4:c.284C>T (p.Pro95Leu)	Missense	27.0
MDS_22	TET2	NM_001127208.2:c.4457C>G; p.(Ser1486*)	Nonsense	41.0

Subject	Gene	Variant	Mutation type	VAF [%]
MDS_22	TET2	NM_001127208.2:c.4138C>T; p.(His1380Tyr)	Missense	35.0
MDS_22	ZRSR2	NM_005089.3:c.491del; p.(Val164Glufs*74)	InDel	72.0
MDS_23	SF3B1	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	82.0
MDS_24	DNMT3A	NM_175629.2:c.2645G>C; p.(Arg882Pro)	Missense	32.0
MDS_24	SF3B1	NM_012433.3:c.1866G>T; p.(Glu622Asp)	Missense	36.0
MDS_24	TET2	NM_001127208.2:c.5618T>C; p.(Ile1873Thr)	Missense	16.0
MDS_25	DNMT3A	NM_175629.2:c.2206C>T; p.(Arg736Cys)	Missense	12.0
MDS_25	RUNX1	NM_001754.4:c.611G>A; p.(Arg204Gln)	Missense	38.0
MDS_25	TET2	NM_001127208.2:c.3415_3418del; p.(Ile1139Leufs*12)	InDel	3.1
MDS_25	WT1	NM_024426.5:c.1453C>T; p.[Arg485Trp]	Missense	40.0
MDS_26	MPL	NM_005373.2:c.1544G>T; p.(Trp515Leu)	Missense	56.0
MDS_26	TET2	NM_001127208.2:c.2713del; p.(Asp905Ilefs*16)	InDel	35.0
MDS_27	RUNX1	NM_001754.4:c.356_358dup; p.(Val119dup)	InDel	7.0
MDS_27	TP53	NM_000546.5:c.814G>A; p.(Val272Met)	Missense	13.0
MDS_27	TP53	NM_000546.5:c.273G>A; p.(Trp91*)	Nonsense	10.0
MDS_28	ASXL1	NM_015338.5:c.1934dup (p.Gly646Trpfs*12)	InDel	23.0
MDS_28	KIT	NM_000222.2:c.2447A>T; p.(Asp816Val)	Missense	12.0
MDS_28	SETBP1	NM_015559.3:c.2608G>A; p.(Gly870Ser)	Missense	6.0
MDS_28	SETBP1	NM_015559.3:c.2602G>A; p.(Asp868Asn)	Missense	3.0
MDS_28	STAT3	NM_139276.2:c.1846G>A; p.(Glu616Lys)	Missense	2.6
MDS_28	TP53	NM_000546.5:c.743G>A; p.(Arg248Gln)	Missense	7.0
MDS_29	DNMT3A	NM_175629.2:c.1093T>A; p.(Tyr365Asn)	Missense	4.0
MDS_29	SRSF2	NM_003016.4:c.284C>T; (p.Pro95Leu)	Missense	1.4
MDS_29	TET2	NM_001127208.2:c.4264A>T; p.(Lys1422*)	Nonsense	2.5
MDS_29	TET2	NM_001127208.2:c.4774dup; p.(Thr1592Asnfs*22)	InDel	2.0
MDS_30	SF3B1	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	45.0
MDS_30	TET2	NM_001127208.2:c.4109G>A; p.(Gly1370Glu)	Missense	30.0
MDS_30	TET2	NM_001127208.2:c.4211_4217del; p.(Arg1404Leufs*42)	InDel	23.0
MDS_31	ASXL1	NM_015338.5:c.1934dup; (p.Gly646Trpfs*12)	InDel	13.0
MDS_31	RUNX1	NM_001754.4:c.602G>A; p.(Arg201Gln)	Missense	28.0
MDS_31	SRSF2	NM_003016.4:c.284C>A; p.(Pro95His)	Missense	25.0
MDS_31	STAG2	NM_001042749.2:c.3034C>T; p.(Arg1012*)	Nonsense	31.0
MDS_32	BCOR	NM_001123385.1:c.2375del; p.(Asn792Ilefs*15)	InDel	4.7
MDS_32	DNMT3A	NM_175629.2:c.2098C>G; p.(Pro700Ala)	Missense	1.1
MDS_33	SF3B1	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	27.0
MDS_34	ASXL2	NM_018263.5:442C>T; p.(Gln148*)	Nonsense	35.0
MDS_34	FLT3	FLT3-ITD (66nt)	InDel	27.0
MDS_34	SF3B1	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	37.0
MDS_84	MPL	NM_005373.2:c.1654-10T>A	Splice site	15.3
MDS_35	DNMT3A	NM_175629.2:c.788_789dup; p.(Pro264Serfs*53)	InDel	33.0
MDS_35	SF3B1	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	37.0
MDS_35	TET2	NM_001127208.2:c.1389_1411dup; p.(Ser471Ilefs*23)	InDel	42.0
MDS_35	TET2	NM_001127208.2:c.2872_2873delCA; p.(Gln958Glufs*13)	InDel	36.0
MDS_35	TET2	NM_001127208.2:c.4075C>A; p.(Arg1359Ser)	Missense	6.3
MDS_36	DNMT3A	NM_175629.2:c.1851+5G>A;p.?	Splice site	3.8

Subject	Gene	Variant	Mutation type	VAF [%]
MDS_36	<i>TP53</i>	NM_000546.5:c.466_477dup; p.(Arg156_Ala159dup)	InDel	3.0
MDS_37	<i>DNMT3A</i>	NM_175629.2:c.1475-1G>C;p.?	Splice site	44.0
MDS_37	<i>SF3B1</i>	NM_012433.3:c.1972T>G; p.(Trp658Gly)	Missense	21.0
MDS_37	<i>SF3B1</i>	NM_012433.3:c.1997A>G; p.(Lys666Arg)	Missense	6.0
MDS_37	<i>SF3B1</i>	NM_012433.3:c.1973G>T; p.(Trp658Leu)	Missense	6.0
MDS_37	<i>SF3B1</i>	NM_012433.3:c.1866G>C; p.(Glu622Asp)	Missense	2.1
MDS_38	<i>RUNX1</i>	NM_001754.4:c.611G>A; p.(Arg204Gln)	Missense	6.0
MDS_38	<i>RUNX1</i>	NM_001754.4:c.602G>A; p.(Arg201Gln)	Missense	2.3
MDS_38	<i>RUNX1</i>	NM_001754.4:c.858_860dup; p.(Tyr287*)	InDel	1.5
MDS_38	<i>SH2B3</i>	NM_005475.2:c.1168delC; p.(Leu390Trpfs*21)	InDel	6.0
MDS_38	<i>SRSF2</i>	NM_003016.4:c.284C>G; p.(Pro95Arg)	Missense	21.0
MDS_38	<i>TET2</i>	NM_001127208.2:c.3646C>T; p.(Arg1216*)	Nonsense	23.0
MDS_38	<i>TET2</i>	NM_001127208.2:c.1454_1455delTG; p.(Val485Glufs*4)	InDel	15.0
MDS_38	<i>TP53</i>	NM_000546.5:c.646G>A; p.(Val216Met)	Missense	7.0
MDS_38	<i>TP53</i>	NM_000546.5:c.743G>A; p.(Arg248Gln)	Missense	2.2
MDS_39	<i>TET2</i>	NM_001127208.2:c.5500_5501delCA; p.(Gln1834Glyfs*11)	InDel	1.9
MDS_40	<i>U2AF1</i>	NM_001025203.1:c.101C>T; p.(Ser34Phe)	Missense	24.0
MDS_41	<i>DDX41</i>	NM_016222.4:c.209dup; p.(Arg71Profs*4)	InDel	39.0
MDS_41	<i>TERT</i>	ENST00000310581:c.3037C>T; p.[His1013Tyr]	Missense	1.3
MDS_42	<i>SF3B1</i>	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	46.4
MDS_42	<i>MPL</i>	NM_005373.2:c.1544G>C; p.(Trp515Ser)	Missense	8.0
MDS_43	<i>ASXL1</i>	NM_015338.5:c.1934dup; (p.Gly646Trpfs*12)	InDel	1.6
MDS_43	<i>DNMT3A</i>	NM_175629.2:c.976C>A; p.(Arg326Ser)	Missense	30.0
MDS_43	<i>DNMT3A</i>	NM_175629.2:c.2644C>T; p.(Arg882Cys)	Missense	1.5
MDS_43	<i>SF3B1</i>	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	32.0
MDS_43	<i>SH2B3</i>	NM_005475.2:c.1090T>C;p.(Trp364Arg)	Missense	2.8
MDS_44	<i>ASXL1</i>	NM_015338.5:c.1934dup; (p.Gly646Trpfs*12)	InDel	19.0
MDS_44	<i>IDH1</i>	NM_005896.3:c.395G>A; p.(Arg132His)	Missense	1.4
MDS_44	<i>IDH2</i>	NM_002168.3:c.419G>A; p.(Arg140Gln)	Missense	27.0
MDS_44	<i>SRSF2</i>	NM_003016.4:c.284C>G; p.(Pro95Arg)	Missense	29.0
MDS_44	<i>STAG2</i>	NM_001042749.2:c.3097C>T; p.(Arg1033*)	Nonsense	36.0
MDS_45	<i>SF3B1</i>	NM_012433.3:c.1997A>G; p.(Lys666Arg)	Missense	24.0
MDS_45	<i>ASXL1</i>	NM_015338.6:c.2827del;p.(Asp943Ilefs*2)	InDel	1.3
MDS_46	<i>DDX41</i>	NM_016222.4:c.1016C>T; p.(Pro339Leu)	Missense	15.0
MDS_47	<i>ASXL1</i>	NM_015338.6:c.1934dup p.(Gly646Trpfs*12)	InDel	13.0
MDS_47	<i>RUNX1</i>	NM_001754.4:c.931_934delACAA;p.(Thr311Profs*16)	InDel	12.3
MDS_48	<i>SF3B1</i>	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	32.0
MDS_49	<i>BCORL1</i>	NM_021946.4:c.[4619-1G>T];p.?	Splice site	83.0
MDS_49	<i>KRAS</i>	NM_033360.3:c.34G>A; p.(Gly12Ser)	Missense	1.1
MDS_49	<i>NRAS</i>	NM_002524.4:c.35G>C; p.(Gly12Ala)	Missense	24.0
MDS_49	<i>NRAS</i>	NM_002524.4:c.37G>C; p.(Gly13Arg)	Missense	7.5
MDS_49	<i>NRAS</i>	NM_002524.4:c.182A>G; p.(Gln61Arg)	Missense	1.1
MDS_49	<i>RUNX1</i>	NM_001754.4:c.664delT; p.(Ser222Profs*15)	InDel	43.0
MDS_49	<i>SF3B1</i>	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	43.0
MDS_49	<i>TET2</i>	NM_001127208.2:c.388delG; p.(Glu130Lysfs*15)	InDel	43.0

Subject	Gene	Variant	Mutation type	VAF [%]
MDS_50	<i>DNMT3A</i>	NM_175629.2:c.2309C>T; p.(Ser770Leu)	Missense	29.0
MDS_50	<i>SF3B1</i>	NM_012433.3:c.1984C>T; p.(His662Tyr)	Missense	33.0
MDS_51	<i>DNMT3A</i>	NM_175629.2: c.2645G>A; p.(Arg882His)	Missense	19.0
MDS_51	<i>DNMT3A</i>	NM_175629.2: c.1667+1G>A;p.?	Splice site	2.3
MDS_51	<i>SF3B1</i>	NM_012433.3:c.2098A>G; p.(Lys700Glu)	Missense	19.0
MDS_51	<i>TET2</i>	NM_001127208.2:c.1691G>A; p.(Trp564*)	Nonsense	2.3
MDS_51	<i>TET2</i>	NM_001127208.2:c.4263C>G; p.(Tyr1421*)	Nonsense	1.4
MDS_52	<i>ASXL1</i>	NM_015338.5:c.2290dup;p.(Leu764Profs*10)	InDel	1.3
MDS_52	<i>CBL</i>	NM_005188.3:c.1202G>C;p.(Cys401Ser)	Missense	1.3
MDS_52	<i>TET2</i>	NM_001127208.2:c.3764dup;p.(Tyr1255*)	InDel	87.0
MDS_53	<i>ASXL1</i>	NM_015338.5:c.1934dup;p.(Gly646Trpfs*12)	InDel	12.0
MDS_53	<i>MPL</i>	NM_005373.2:c.1775G>A;p.(Arg592Gln)	Missense	15.0
MDS_53	<i>U2AF1</i>	NM_001025203.1:c.470A>C;p.(Gln157Pro)	Missense	21.0
MDS_54	<i>ASXL1</i>	NM_015338.5:c.1819_1820delGG;p.(Gly607Leufs*11)	InDel	14.0
MDS_54	<i>SF3B1</i>	NM_012433.3:c.2342A>G;p.(Asp781Gly)	Missense	33.0
MDS_54	<i>TP53</i>	NM_000546.5:c.818G>A;p.(Arg273His)	Missense	34.0
MDS_55	<i>DNMT3A</i>	NM_175629.2:c.1429+1G>A;p.?	Splice site	1.5
MDS_56	<i>DNMT3A</i>	NM_175629.2:c.2645G>A;p.(Arg882His)	Missense	3.7
MDS_56	<i>SRSF2</i>	NM_003016.4:c.284_307delCCCCGGACTCACACCACAGCCGCC;p.(Pro95_Arg102del)	InDel	15.0
MDS_56	<i>TET2</i>	NM_001127208.2:c.3894dup;p.(Lys1299*)	InDel	16.0
MDS_57	<i>ASXL1</i>	NM_015338.5:c.2324T>G;p.(Leu775*)	Nonsense	1.1
MDS_57	<i>SF3B1</i>	NM_012433.3:c.1874G>T;p.(Arg625Leu)	Missense	6.4
MDS_57	<i>TET2</i>	NM_001127208.2:c.3658delA;p.(Thr1220Profs*6)	InDel	7.7
MDS_58	<i>RAD21</i>	NM_006265.3:c.1064C>T;p.(Pro355Leu)	Missense	21.4
MDS_58	<i>SRSF2</i>	NM_003016.4:c.284_307delCCCCGGACTCACACCACAGCCGCC;p.(Pro95_Arg102del)	InDel	23.4
MDS_58	<i>TET2</i>	NM_001127208.2:c.4044+1G>C;p.?	Splice site	10.0
MDS_59	<i>DNMT3A</i>	NM_175629.2:c.2612delC;p.(Pro871Glnfs*10)	InDel	3.7
MDS_59	<i>SF3B1</i>	NM_012433.3:c.1868A>G;p.(Tyr623Cys)	Missense	24.0
MDS_59	<i>SRSF2</i>	NM_003016.4:c.[283C>G];p.(Pro95Ala)	Missense	3.8
MDS_59	<i>TET2</i>	NM_001127208.2:c.3862G>C;p.(Gly1288Arg)	Missense	2.2
MDS_60	<i>RUNX1</i>	NM_001754.4:c.292delC;p.(Leu98Serfs*24)	InDel	1.7
MDS_60	<i>SRSF2</i>	NM_003016.4:c.284_307delCCCCGGACTCACACCACAGCCGCC;p.(Pro95_Arg102del)	InDel	26.0
MDS_60	<i>TET2</i>	NM_001127208.2:c.3921delG;p.(Lys1308Serfs*55)	InDel	1.8
MDS_60	<i>TET2</i>	NM_001127208.2:c.327delC;p.(Lys110Argfs*3)	InDel	4.0
MDS_61	<i>ASXL1</i>	NM_015338.5:c.1934dup;p.(Gly646Trpfs*12)	InDel	30.0
MDS_61	<i>BRAF</i>	NM_001354609.2:c.602A>G;p.(Gln201Arg)	Missense	44.0
MDS_61	<i>EZH2</i>	NM_004456.5:c.324delC;p.(Ile109*)	InDel	51.0
MDS_61	<i>FLT3</i>	NM_004119.3:c.2523C>A;p.(Asn841Lys)	Missense	40.0
MDS_61	<i>GATA2</i>	NM_001145661.2:c.229+1G>T;p.?	Splice site	40.0
MDS_61	<i>RUNX1</i>	NM_001754.4:c.[1207_1210dup]; p.(His404Leufs*197)	InDel	4.2
MDS_61	<i>RUNX1</i>	NM_001754.4:c.[951_952delTT];p.(Ser318Glnfs*281)	InDel	34.0
MDS_62	<i>ASXL1</i>	NM_015338.5:c.1934dup;p.(Gly646Trpfs*12)	InDel	12.3
MDS_62	<i>CEBPA</i>	NM_004364:c.514C>T;p.(Gln172*)	Nonsense	19.0

Subject	Gene	Variant	Mutation type	VAF [%]
MDS_62	<i>SF3B1</i>	NM_012433.3:c.1873C>T;p.(Arg625Cys)	Missense	15.0
MDS_62	<i>STAG2</i>	NM_001042750.2STAG2:c.1907dup;p.(Tyr636*)	InDel	30.0
MDS_62	<i>TET2</i>	NM_001127208.2:c.1813_1814delTA;p.(Tyr605Hisfs*32)	InDel	8.4
MDS_62	<i>TET2</i>	NM_001127208.2:c.5456delT;p.(Leu1819*)	InDel	18.0
MDS_63	<i>CBL</i>	NM_005188.3:c.1222T>G;p.(Trp408Gly)	Missense	16.3
MDS_63	<i>MPL</i>	NM_005373.3:c.545C>A; p.(Pro182His)	Missense	2.9
MDS_63	<i>NOTCH1</i>	ENST00000277541:c.7541_7542delCT;p.Pro2514fs	InDel	1.6
MDS_63	<i>TET2</i>	NM_001127208.2:c.4029_4030insC; p.(Ala1344Argfs*3)	InDel	34.6
MDS_64	<i>CBL</i>	NM_005188.3:c.1256G>A;p.(Cys419Tyr)	Missense	11.0
MDS_64	<i>EZH2</i>	NM_004456.5:c.2198A>G;p.(Tyr733Cys)	Missense	35.9
MDS_64	<i>TET2</i>	NM_001127208.2:c.5269delC;p.(His1757Ilefs*6)	InDel	19.7
MDS_65	<i>CSF3R</i>	NM_156039.3:c.2302C>T;p.(Gln768*)	Nonsense	2.5
MDS_65	<i>CSF3R</i>	NM_156039.3:c.2337T>A; p.(Tyr779*)	Nonsense	24.2
MDS_65	<i>DNMT3A</i>	NM_175629.2:c.2645G>A p.(Arg882His)	Missense	27.8
MDS_65	<i>U2AF1</i>	NM_001025203.1:c.470A>G;p.(Gln157Arg)	Missense	23.7
MDS_66	<i>TP53</i>	NM_000546.5:c.427G>A; p.(Val143Met)	Missense	27.4
MDS_67	<i>IDH1</i>	NM_005896.3: c.394C>T p.(Arg132Cys)	Missense	3.5
MDS_67	<i>RUNX1</i>	NM_001754.4:c.293T>C; p.(Leu98Pro)	Missense	30.3
MDS_67	<i>U2AF1</i>	NM_001025203.1:c.470A>C p.(Gln157Pro)	Missense	36.1
MDS_68	<i>SF3B1</i>	NM_012433.3:c.2098A>G p.(Lys700Glu)	Missense	39.0
MDS_69	<i>DNMT3A</i>	NM_175629.2:c.1851+1G>A; p.?	Splice site	4.7
MDS_69	<i>TET2</i>	NM_001127208.2:c.822del;p.(Asn275Ilefs*18)	InDel	3.1
MDS_69	<i>TP53</i>	NM_000546.5:c.473G>A;p.(Arg158His)	Missense	55.2
MDS_70	<i>ASXL1</i>	NM_015338.6:c.2535dup;p.(Ser846Glnfs*5)	InDel	33.0
MDS_70	<i>ETNK1</i>	NM_018638.4:c.731A>G;p.(Asn244Ser)	Missense	7.8
MDS_70	<i>U2AF1</i>	NM_001025203.1:c.470A>C p.(Gln157Pro)	Missense	6.9
MDS_70	<i>CALR</i>	NM_004343.3:c.1099_1150del;p.(Leu367Thrfs*46)	InDel	1.8
MDS_71	<i>ASXL1</i>	NM_015338.6:c.1804G>T;p.(Glu602*)	Nonsense	39.8
MDS_71	<i>CBL</i>	NM_005188.3:c.1151G>T;p.(Cys384Phe)	Missense	13.4
MDS_71	<i>ETNK1</i>	NM_018638.4:c.731A>G;p.(Asn244Ser)	Missense	1.7
MDS_71	<i>SETBP1</i>	NM_015559.3:c.2608G>A (p.Gly870Ser)	Missense	40.7
MDS_71	<i>SRSF2</i>	NM_003016.4:c.284C>T p.(Pro95Leu)	Missense	27.9
MDS_71	<i>ZRSR2</i>	NM_005089.3:c.1318_1319insCGGCC;p.(Arg440Profs*?)	InDel	36.0
MDS_72	<i>DNMT3A</i>	NM_175629.2:c.2628C>A;p.(Asp876Glu)	Missense	38.3
MDS_72	<i>DNMT3A</i>	NM_175629.2:c.2408+1G>A;p.?	Splice site	40.5
MDS_72	<i>SF3B1</i>	NM_012433.3:c.2098A>G p.(Lys700Glu)	Missense	40.9
MDS_72	<i>TET2</i>	NM_001127208.2:c.4115C>T;p.(Thr1372Ile)	Missense	39.8
MDS_73	<i>JAK2</i>	NM_004972.3:c.1849G>T p.(Val617Phe)	Missense	31.4
MDS_73	<i>TET2</i>	NM_001127208.2:c.2899C>T;p.(Gln967*)	Nonsense	40.5
MDS_73	<i>TET2</i>	NM_001127208.2:c.3594+4A>G;p.?	Splice site	42.3
MDS_73	<i>ZRSR2</i>	NM_005089.3:c.959G>A;p.(Cys320Tyr)	Missense	81.6
MDS_74	<i>EZH2</i>	NM_004456.5:c.2006G>A;p.(Ser669Asn)	Missense	83.7
MDS_74	<i>RUNX1</i>	NM_001754.4:c.602G>A;p.(Arg201Gln)	Missense	5.0
MDS_74	<i>SETBP1</i>	NM_015559.3:c.2602G>A p.(Asp868Asn)	Missense	31.7
MDS_74	<i>SF3B1</i>	NM_012433.3:c.1986C>G;p.(His662Gln)	Missense	42.7

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MDS_75	<i>DDX41</i>	NM_016222.2:c.1127C>T;p.(Ala376Val)	Missense	6.0
MDS_75	<i>DNMT3A</i>	NM_175629.2:c.2723A>C;p.(Tyr908Ser)	Missense	11.9
MDS_75	<i>TET2</i>	NM_001127208.2:c.3865T>G;p.(Cys1289Gly)	Missense	2.0
MDS_76	<i>DNMT3A</i>	NM_175629.2: c.2645G>A p.(Arg882His)	Missense	14.3
MDS_76	<i>DNMT3A</i>	NM_175629.2:c.2327A>G;p.(Asn776Ser)	Missense	17.9
MDS_77	<i>SF3B1</i>	NM_012433.3:c.2098A>G p.(Lys700Glu)	Missense	5.0
MDS_77	<i>RAD21</i>	NM_006265.3:c.415_419del;p.(Arg139Glyfs*19)	InDel	4.6
MDS_78	<i>TET2</i>	NM_001127208.2:c.1397del;p.(Ser466Leufs*20)	InDel	2.2
MDS_78	<i>TET2</i>	NM_001127208.2:c.4546C>T;p.(Arg1516*)	Nonsense	20.3
MDS_78	<i>TET2</i>	NM_001127208.2:c.3410-4A>G;p.?	Splice site	43.8
MDS_78	<i>DNMT3A</i>	NM_175629.2:c.1302del;p.(Val435Cysfs*216)	InDel	44.9
MDS_78	<i>SH2B3</i>	NM_005475.2:c.1153G>C;p.(Ala385Pro)	Missense	1.9
MDS_78	<i>JAK2</i>	NM_004972.3:c.1849G>T;p.(Val617Phe)	Missense	2.5
MDS_79	<i>TP53</i>	NM_000546.5:c.559+1G>T;p.?	Splice site	36.5
MDS_80	<i>SRSF2</i>	NM_003016.4:c.284C>A p.(Pro95His)	Missense	35.2
MDS_80	<i>ASXL1</i>	NM_015338.6:c.1904_1905del;p.(Glu635Glyfs*22)	InDel	37.6
MDS_80	<i>RUNX1</i>	NM_001754.4:c.998dup;p.(Arg334Alafs*266)	InDel	33.8
MDS_80	<i>KRAS</i>	NM_033360.4:c.437C>T p.(Ala146Val)	Missense	4.0
MDS_81	<i>JAK2</i>	NM_004972.3:c.1849G>T p.(Val617Phe)	Missense	5.0
MDS_81	<i>U2AF1</i>	NM_001025203.1:c.470A>G p.(Gln157Arg)	Missense	11.0
MDS_82	<i>TET2</i>	NM_001127208.2:c.3671del;p.(Ala1224Glyfs*2)	InDel	1.8
MDS_82	<i>TP53</i>	NM_000546.5:c.659A>G;p.(Tyr220Cys)	Missense	18.5
MDS_82	<i>BRAF</i>	NM_004333.4:c.1406G>C;p.(Gly469Ala)	Missense	1.1
MDS_82	<i>ZBTB7A</i>	NM_015898.4:c.231del;p.(Ile77Metfs*43)	InDel	5.3
MDS_83	<i>SRSF2</i>	NM_003016.4:c.284C>A p.(Pro95His)	Missense	46.6
MDS_83	<i>ASXL1</i>	NM_015338.6:c.2182G>T;p.(Glu728*)	Nonsense	41.0
MDS_83	<i>STAG2</i>	NM_001042750.2:c.1840C>T p.(Arg614*)	Nonsense	86.0
AML_1	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG;p.G642fs	InDel	11.6
AML_1	<i>BCOR</i>	NM_001123385:exon7:c.3446_3447del;p.A1149fs	InDel	63.9
AML_1	<i>BCOR</i>	NM_001123385:exon7:c.3444_3446G	InDel	35.2
AML_1	<i>CSF3R</i>	NM_156039:exon17:c.2436_2437insGCAG;p.S813fs	InDel	1.2
AML_1	<i>IDH2</i>	NM_002168:exon4:c.419G>A;p.R140Q	Missense	32.7
AML_1	<i>NRAS</i>	NM_002524:exon2:c.35G>T;p.G12V	Missense	18.9
AML_1	<i>PHF6</i>	NM_032458:exon9:c.954dupA;p.S318fs	InDel	5.9
AML_1	<i>RUNX1</i>	NM_001754:exon9:c.1141_1142insCGCC;p.L381fs	InDel	31.8
AML_1	<i>SRSF2</i>	NM_003016:exon1:c.284C>A;p.P95H	Missense	2.5
AML_1	<i>STAG2</i>	NM_001042750:exon15:c.1409delA;p.E470fs	InDel	61.0
AML_1	<i>TET2</i>	NM_001127208:exon3:c.2692G>T;p.G898X	Nonsense	4.0
AML_2	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG;p.G642fs	InDel	18.4
AML_2	<i>CEBPA</i>	NM_004364:exon1:c.898C>T;p.R300C	Missense	25.1
AML_2	<i>RUNX1</i>	NM_001754:exon4:c.317G>T;p.W106L	Missense	33.8
AML_2	<i>STAG2</i>	NM_001042750:exon24:c.2308C>T;p.Q770X	Nonsense	27.6
AML_2	<i>U2AF1</i>	NM_001025203:exon6:c.470A>C;p.Q157P	Missense	28.8
AML_3	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG;p.G642fs	InDel	15.2
AML_3	<i>BRAF</i>	NM_004333:c.1790T>A;p.L597Q	Missense	90.5

Subject	Gene	Variant	Mutation type	VAF [%]
AML_3	<i>EZH2</i>	NM_004456:exon19:c.2186T>C:p.F729S	Missense	92.3
AML_3	<i>IDH2</i>	NM_002168:exon4:c.419G>A:p.R140Q	Missense	46.4
AML_3	<i>RUNX1</i>	NM_001754:exon4:c.329A>C:p.K110T	Missense	3.1
AML_3	<i>STAG2</i>	NM_001042750:exon20:c.1840C>T:p.R614X	Nonsense	5.5
AML_3	<i>STAG2</i>	NM_001042750:exon21:c.2086_2096del:p.A696fs	InDel	7.9
AML_3	<i>STAG2</i>	NM_001042750:exon31:c.3394_3395insTA:p.L1132fs	InDel	27.9
AML_4	<i>ASXL1</i>	NM_015338:exon12:c.2077C>T:p.R693X	Nonsense	8.6
AML_4	<i>KRAS</i>	NM_033360:exon2:c.35G>A:p.G12D	Missense	38.3
AML_5	<i>FLT3</i>	NM_004119:exon20:c.2508_2510del:p.836_837del	InDel	7.2
AML_5	<i>U2AF1</i>	NM_001025203:exon2:c.101C>T:p.S34F	Missense	50.9
AML_6	<i>ASXL1</i>	NM_015338:exon12:c.2160delC:p.D720fs	InDel	42.9
AML_6	<i>RUNX1</i>	NM_001754:exon6:c.610C>T:p.R204X	Nonsense	38.6
AML_6	<i>SRSF2</i>	NM_003016:exon1:c.284C>A:p.P95H	Missense	41.9
AML_6	<i>TET2</i>	NM_001127208:exon3:c.757_770del:p.N253fs	InDel	40.0
AML_7	<i>IDH1</i>	NM_005896:exon4:c.394C>T:p.R132C	Missense	44.6
AML_7	<i>RUNX1</i>	NM_001754:exon5:c.369dupT:p.V124fs	InDel	47.2
AML_7	<i>SRSF2</i>	NM_003016:exon1:c.284C>T:p.P95L	Missense	40.4
AML_7	<i>TET2</i>	NM_001127208:exon11:c.5482C>T:p.Q1828X	Nonsense	99.0
AML_8	<i>BCOR</i>	NM_001123385:exon11:c.4428+2T>G,p.?	Splice site	50.8
AML_8	<i>BCORL1</i>	NM_021946:exon3:c.398_399insCGGT:p.S133fs	InDel	50.2
AML_8	<i>DNMT3A</i>	NM_175629:exon23:c.2644C>T:p.R882C	Missense	56.4
AML_8	<i>KIT</i>	NM_000222:exon8:c.1256_1261del:p.419_421del	InDel	41.3
AML_8	<i>RUNX1</i>	NM_001754:exon9:c.1007_1008insAGGGCCC:p.F336fs	InDel	41.0
AML_8	<i>SF3B1</i>	NM_012433:exon15:c.2098A>G:p.K700E	Missense	48.0
AML_9	<i>DNMT3A</i>	NM_175629:exon23:c.2645G>A:p.R882H	Missense	34.5
AML_9	<i>IDH2</i>	NM_002168:exon4:c.419G>A:p.R140Q	Missense	39.5
AML_9	<i>RUNX1</i>	NM_001754:exon9:c.1015_1016insCAGTTCCCCGCGC:p.L339fs	InDel	27.1
AML_10	<i>ETV6</i>	NM_001987:exon3:c.313_314insT:p.R105fs	InDel	37.7
AML_10	<i>FLT3</i>	NM_004119:exon14:c.1769_1770insTCAGCCCATCGGCTCCTCAGATAATGAGTACTT:p.F590delinsFQPIGSSDNEYF	InDel	25.3
AML_10	<i>NPM1</i>	NM_002520:exon10:c.805_806insGGGCGC:p.I269delinsRAL	InDel	37.9
AML_11	<i>DNMT3A</i>	NM_175629:exon13:c.1474+1G>A,p.?	Splice site	46.5
AML_11	<i>FLT3</i>	NM_004119:exon14:c.1799_1800insGTCTCCTCCTCAGATAATGAGTACTTCTACGTTGATTTTCAGAGAATATGAATATGA:p.D600delinsESSDNEYFYVDFREYED	InDel	1.2
AML_11	<i>RUNX1</i>	NM_001754:exon5:c.356dupT:p.V119fs	InDel	29.0
AML_11	<i>RUNX1</i>	NM_001754:exon4:c.299_300insATCGCAGAGGAGCAA:p.S100delinsSSQRSN	InDel	9.5
AML_11	<i>STAG2</i>	NM_001042750:exon28:c.2857C>T:p.R953X	Nonsense	26.2
AML_11	<i>U2AF1</i>	NM_001025203:exon2:c.101C>A:p.S34Y	Missense	52.4
AML_12	<i>ASXL1</i>	NM_015338:exon12:c.1888_1910del:p.H630fs	InDel	33.9
AML_12	<i>IDH2</i>	NM_002168:exon4:c.419G>A:p.R140Q	Missense	47.0
AML_12	<i>SRSF2</i>	NM_003016:exon1:c.284C>A:p.P95H	Missense	42.6
AML_12	<i>STAG2</i>	NM_001042750:exon20:c.1840C>T:p.R614X	Nonsense	14.6
AML_12	<i>STAG2</i>	NM_001042750:exon29:c.3034C>T:p.R1012X	Nonsense	65.2
AML_13	<i>NRAS</i>	NM_002524:exon2:c.38G>A:p.G13D	Missense	39.8

Subject	Gene	Variant	Mutation type	VAF [%]
AML_13	SF3B1	NM_012433:exon15:c.2098A>G;p.K700E	Missense	32.0
AML_14	ASXL1	NM_015338:exon12:c.1888_1910del:p.630_637del	InDel	43.7
AML_14	BCOR	NM_001123385:exon7:c.3490C>T;p.R1164X	Nonsense	94.2
AML_14	DNMT3A	NM_175629:exon18:c.2163G>C;p.K721N	Missense	89.3
AML_14	ETV6	NM_001987:exon3:c.164-2A>G,p.?	Splice site	44.5
AML_14	NRAS	NM_002524:exon2:c.35G>A;p.G12D	Missense	53.1
AML_14	PTPN11	NM_002834:exon3:c.181G>T;p.D61Y	Missense	12.9
AML_14	RUNX1	NM_001754:exon7:c.664delT;p.S222fs	InDel	39.5
AML_15	KIT	NM_000222:exon17:c.2447A>T;p.D816V	Missense	38.3
AML_15	NRAS	NM_002524:exon3:c.183A>C;p.Q61H	Missense	5.7
AML_15	NRAS	NM_002524:exon3:c.182A>G;p.Q61R	Missense	21.0
AML_15	PTPN11	NM_002834:exon3:c.226G>A;p.E76K	Missense	14.7
AML_15	SF3B1	NM_012433:exon15:c.2098A>G;p.K700E	Missense	44.7
AML_16	CEBPA	NM_004364:exon1:c.175G>T;p.E59X	Nonsense	76.7
AML_16	CSF3R	NM_156039:exon15:c.1919C>A;p.T640N	Missense	18.4
AML_16	RUNX1	NM_001754:exon9:c.1354G>A;p.V452M	Missense	57.5
AML_16	SRSF2	NM_003016:exon1:c.284C>A;p.P95H	Missense	41.6
AML_16	STAG2	NM_001042750:exon11:c.1015delA;p.K339fs	InDel	28.5
AML_16	TET2	NM_001127208:exon3:c.3315_3316del:p.I1105fs	InDel	44.1
AML_16	TET2	NM_001127208:exon6:c.3796A>G;p.N1266D	Missense	49.1
AML_16	WT1	NM_024426:exon7:c.1099-1G>A,p.?	Splice site	12.5
AML_17	ASXL1	NM_015338:exon12:c.2058_2059del:p.K686fs	InDel	45.3
AML_17	BCOR	NM_001123385:exon11:c.4540C>T;p.R1514X	Nonsense	38.9
AML_17	NRAS	NM_002524:exon2:c.35G>A;p.G12D	Missense	48.0
AML_17	RUNX1	NM_001754:exon4:c.245_246insAGCA;p.L82fs	InDel	56.0
AML_17	SRSF2	NM_003016:exon1:c.284C>A;p.P95H	Missense	44.4
AML_17	STAG2	NM_001042750:exon9:c.775C>T;p.R259X	Nonsense	45.1
AML_17	TET2	NM_001127208:exon5:c.3589A>G;p.K1197E	Missense	47.6
AML_18	ASXL1	NM_015338:exon12:c.1888_1910del:p.630_637del	InDel	25.8
AML_18	IDH2	NM_002168:exon4:c.515G>A;p.R172K	Missense	21.7
AML_18	PTPN11	NM_002834:exon13:c.1517A>C;p.Q506P	Missense	14.4
AML_18	RUNX1	NM_001754:exon6:c.610C>T;p.R204X	Nonsense	36.9
AML_18	RUNX1	NM_001754:exon6:c.592G>A;p.D198N	Missense	4.1
AML_18	SRSF2	NM_003016:exon1:c.284C>A;p.P95H	Missense	37.4
AML_18	TET2	NM_001127208:exon11:c.4834_4835del:p.1612_1612del	InDel	40.8
AML_19	ASXL1	NM_015338:exon12:c.2084delA;p.Q695fs	InDel	2.0
AML_19	SRSF2	NM_003016:exon1:c.284C>A;p.P95H	Missense	44.9
AML_19	TET2	NM_001127208:exon3:c.2218C>T;p.Q740X	Nonsense	48.3
AML_19	TET2	NM_001127208:exon7:c.3863G>A;p.G1288D	Missense	44.5
AML_20	IDH2	NM_002168:exon4:c.419G>A;p.R140Q	Missense	44.0
AML_21	DNMT3A	NM_175629:exon23:c.2645G>A;p.R882H	Missense	48.6
AML_22	DNMT3A	NM_175629:exon23:c.2644C>T;p.R882C	Missense	45.8
AML_22	FLT3	NM_004119:exon20:c.2503G>T;p.D835Y	Missense	7.6
AML_22	RUNX1	NM_001754:exon9:c.978_979insG;p.L327fs	InDel	46.1
AML_22	SF3B1	NM_012433:exon15:c.2098A>G;p.K700E	Missense	49.8

Subject	Gene	Variant	Mutation type	VAF [%]
AML_23	GATA2	NM_001145661:exon7:c.1160_1171del:p.387_391del	InDel	33.7
AML_23	NRAS	NM_002524:exon3:c.181C>A:p.Q61K	Missense	39.8
AML_24	NPM1	NM_002520:exon11:c.861_862insTGCA:p.L287fs	InDel	27.5
AML_24	PTPN11	NM_002834:exon3:c.179G>T:p.G60V	Missense	2.3
AML_24	RAD21	NM_006265:exon6:c.640C>T:p.Q214X	Nonsense	2.6
AML_25	ASXL2	NM_018263:exon11:c.1771_1772insCCCCACTGAGAATC:p.R591fs	InDel	47.2
AML_25	FLT3	NM_004119:exon15:c.1837+2->CGTGTTTCAGAGAATATGAATATGATCTCAAATGGGAGTTTCCAAGAGAAAATTTAGAGTTTGG	Splice site	13.7
AML_25	FLT3	NM_004119:exon14:c.1790_1791insCTCACCTAACGATTTCA GAGAATA:p.Y597delinsYSPNDFREY	InDel	20.1
AML_25	RUNX1	NM_001754:exon8:c.861C>G:p.Y287X	Nonsense	45.0
AML_25	SF3B1	NM_012433:exon14:c.1996A>C:p.K666Q	Missense	38.4
AML_25	WT1	NM_024426:exon7:c.1137_1138insGAGGCGGGTC:p.R380fs	InDel	7.8
AML_26	DNMT3A	NM_175629:exon23:c.2645G>C:p.R882P	Missense	33.3
AML_26	FLT3	NM_004119:exon13:c.1613_1805GAGAATATGAATATGATCTCAAATGGGAGTT	InDel	15.7
AML_26	FLT3	NM_004119:exon14:c.1802_1803insTGATTTTCAGAGAATATG AATATGATCT:p.L601delinsLDFREYDYDL	InDel	4.1
AML_26	FLT3	NM_004119:exon14:c.1800_1801insTTCAGAGAATATGAATA TGAT:p.L601delinsFREYDYDL	InDel	23.9
AML_26	NPM1	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	38.4
AML_27	KRAS	NM_033360:exon2:c.38G>A:p.G13D	Missense	20.7
AML_27	NPM1	NM_002520:exon11:c.861_862insTGCA:p.L287fs	InDel	46.2
AML_28	MPL	NM_005373:exon10:c.1544G>T:p.W515L	Missense	2.2
AML_28	U2AF1	NM_001025203:exon6:c.470A>C:p.Q157P	Missense	20.4
AML_29	DDX41	NM_016222:exon15:c.1574G>A:p.R525H	Missense	7.8
AML_29	TET2	NM_001127208:exon7:c.3894T>A:p.C1298X	Nonsense	16.1
AML_30	ETV6	NM_001987:exon5:c.976_977insTTGGACCT:p.E326fs	InDel	0.6
AML_30	WT1	NM_024426:exon7:c.1138_1139insA:p.R380fs	InDel	22.5
AML_31	BCOR	NM_001123385:exon4:c.1005dupC:p.S336fs	InDel	44.3
AML_31	BCORL1	NM_021946:c.3001C>T:p.Q1001X	Nonsense	46.3
AML_31	KRAS	NM_033360:exon2:c.35G>A:p.G12D	Missense	42.4
AML_31	RUNX1	NM_001754:exon5:c.484A>G:p.R162G	Missense	27.9
AML_32	CEBPA	NM_004364:exon1:c.1000G>A:p.E334K	Missense	43.1
AML_32	DNMT3A	NM_175629:exon23:c.2645G>A:p.R882H	Missense	35.8
AML_32	KRAS	NM_033360:exon3:c.183A>T:p.Q61H	Missense	31.5
AML_32	NPM1	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	40.2
AML_32	PTPN11	NM_002834:exon3:c.182A>G:p.D61G	Missense	4.5
AML_32	RAD21	NM_006265:exon2:c.59C>T:p.A20V	Missense	3.7
AML_33	BCOR	NM_001123385:exon11:c.4428+1G>A,p.?	Splice site	86.6
AML_33	KRAS	NM_033360:exon2:c.37G>C:p.G13R	Missense	28.4
AML_33	NRAS	NM_002524:exon2:c.34G>A:p.G12S	Missense	12.4
AML_33	RUNX1	NM_001754:exon5:c.455dupA:p.K152fs	InDel	43.5
AML_33	STAG2	NM_001042750:exon20:c.1907_1908insTCCTC:p.Y636fs	InDel	85.4
AML_33	TET2	NM_001127208:exon11:c.5587G>A:p.A1863T	Missense	46.7
AML_34	DNMT3A	NM_175629:exon23:c.2644C>T:p.R882C	Missense	42.9

Subject	Gene	Variant	Mutation type	VAF [%]
AML_34	DNMT3A	NM_175629:exon18:c.2173G>A:p.E725K	Missense	42.4
AML_34	JAK2	NM_004972:exon14:c.1849G>T:p.V617F	Missense	14.9
AML_34	SRSF2	NM_003016:exon1:c.284C>A:p.P95H	Missense	38.1
AML_34	TET2	NM_001127208:exon7:c.3894dupT:p.C1298fs	InDel	81.9
AML_35	NPM1	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	36.8
AML_35	STAG2	NM_001042750:p.?	Splice site	70.8
AML_36	TP53	NM_000546:exon7:c.742C>T:p.R248W	Missense	96.3
AML_37	BCOR	NM_001123385:exon4:c.2962_2963insGT:p.S988fs	InDel	67.9
AML_37	IDH2	NM_002168:exon4:c.419G>A:p.R140Q	Missense	46.0
AML_37	SRSF2	NM_003016:exon1:c.284_307del:p.95_103del	InDel	41.2
AML_38	BCOR	NM_001123385:exon10:c.4425T>A:p.Y1475X	Nonsense	42.9
AML_38	DNMT3A	NM_175629:exon22:c.2510C>A:p.S837X	Nonsense	50.2
AML_38	DNMT3A	NM_175629:exon13:c.1490G>C:p.C497S	Missense	36.9
AML_38	NPM1	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	37.6
AML_38	SF3B1	NM_012433:exon15:c.2098A>G:p.K700E	Missense	42.8
AML_38	TET2	NM_001127208:exon3:c.2101C>T:p.Q701X	Nonsense	50.7
AML_38	TET2	NM_001127208:exon3:c.3249_3250A	InDel	2.7
AML_38	TET2	NM_001127208:exon10:c.4468G>T:p.E1490X	Nonsense	42.2
AML_39	NRAS	NM_002524:exon3:c.181C>A:p.Q61K	Missense	43.0
AML_39	SF3B1	NM_012433:exon15:c.2098A>G:p.K700E	Missense	49.5
AML_39	WT1	NM_024426:exon7:c.1144_1145insTCGG:p.A382fs	InDel	42.6
AML_40	FLT3	NM_004119:exon14:c.1809_1810insAGAGAATATGAATATGATCTCAAATGG:p.E604delinsREYEYDLKWE	InDel	8.9
AML_40	FLT3	NM_004119:exon14:c.1787_1788insCTTCTACGTTGATTTCA GAGA:p.E596delinsDFYVDFRE	InDel	14.4
AML_40	FLT3	NM_004119:exon14:c.1787_1788insGATGGTACAGGTGACC GGCTCCTCAGATAATGAGTACTTCTACGTTGATTTTCAGAG A:p.E596delinsEMVQVTGSSDNEYFYVDFRE	InDel	24.4
AML_41	SF3B1	NM_012433:exon15:c.2098A>G:p.K700E	Missense	44.6
AML_41	TP53	NM_000546:exon4:c.325T>G:p.F109V	Missense	82.5
AML_41	WT1	NM_024426:exon1:c.332dupC:p.P111fs	InDel	46.0
AML_42	BCOR	NM_001123385:exon4:c.1882G>A:p.A628T	Missense	2.3
AML_42	BCOR	NM_001123385:exon4:c.1240G>A:p.A414T	Missense	3.2
AML_42	FLT3	NM_004119:exon15:c.1837+3->GATTTTCAGAGAATATGAATATGATCTCAAATGGGAGTTTC CAAGAGAAAATTTAGAGTTTGGT	Splice site	37.3
AML_42	STAG2	NM_001042750:exon32:c.3468-3A>G,p.?	Splice site	2.1
AML_42	TP53	NM_000546:exon10:c.1004G>A:p.R335H	Missense	2.4
AML_42	WT1	NM_024426:exon7:c.1144_1145insTCGG:p.A382fs	InDel	43.5
AML_43	PTPN11	NM_002834:exon3:c.227A>G:p.E76G	Missense	21.7
AML_43	RUNX1	NM_001754:exon5:c.496C>T:p.R166X	Nonsense	37.1
AML_44	CSF3R	NM_156039:exon14:c.1853C>T:p.T618I	Missense	41.8
AML_44	GATA2	NM_001145661:exon7:c.1187G>T:p.R396L	Missense	39.4
AML_44	NRAS	NM_002524:exon3:c.182A>G:p.Q61R	Missense	16.2
AML_44	SETBP1	NM_015559:exon4:c.2608G>A:p.G870S	Missense	46.5
AML_44	U2AF1	NM_001025203:exon6:c.470A>C:p.Q157P	Missense	46.7

Subject	Gene	Variant	Mutation type	VAF [%]
AML_44	WT1	NM_024426:exon2:c.749dupT:p.M250fs	InDel	78.7
AML_45	ASXL1	NM_015338:exon12:c.1926dupA:p.G642fs	InDel	31.1
AML_45	ETV6	NM_001987:exon4:c.329-2A>G,p.?	Splice site	49.0
AML_45	RUNX1	NM_001754:exon7:c.676_677del:p.S226fs	InDel	46.7
AML_45	SRSF2	NM_003016:exon1:c.284C>G:p.P95R	Missense	49.0
AML_45	TET2	NM_001127208:exon3:c.2490dupA:p.I830fs	InDel	37.9
AML_46	ASXL1	NM_015338:exon12:c.1888_1910del:p.H630fs	InDel	36.1
AML_46	FLT3	NM_004119:exon14:c.1769_1770insGCCTCAGATAACCGGC TCCTCAGATAATGAGTACTT:p.F590delinsLPQITGSSDNEYF	InDel	69.4
AML_46	GATA2	NM_001145661:exon4:c.416_417del:p.S139fs	InDel	41.2
AML_46	PTPN11	NM_002834:exon3:c.215C>A:p.A72D	Missense	4.8
AML_46	RUNX1	NM_001754:exon7:c.623dupA:p.Q208fs	InDel	34.6
AML_46	SRSF2	NM_003016:exon1:c.284C>A:p.P95H	Missense	41.0
AML_46	STAG2	NM_001042750:exon13:c.1117-1->GATA,p.?	Splice site	12.5
AML_46	STAG2	NM_001042750:exon28:c.2857C>T:p.R953X	Nonsense	8.7
AML_46	STAG2	NM_001042750:exon28:c.2915dupT:p.M972fs	InDel	69.6
AML_47	DDX41	NM_016222:exon2:c.121C>T:p.Q41X	Nonsense	48.8
AML_48	BCOR	NM_001123385:exon12:c.4647_4649del:p.1549_1550del	InDel	17.9
AML_48	IDH1	NM_005896:exon4:c.394C>T:p.R132C	Missense	8.3
AML_48	RUNX1	NM_001754:p.?	Splice site	13.2
AML_48	SRSF2	NM_003016:exon1:c.284C>A:p.P95H	Missense	13.0
AML_48	STAG2	NM_001042750:exon10:c.868dupA:p.F289fs	InDel	7.5
AML_48	WT1	NM_024426:exon10:c.1462A>G:p.S488G	Missense	52.7
AML_49	DNMT3A	NM_175629:exon8:c.958C>T:p.R320X	Nonsense	32.9
AML_49	IDH1	NM_005896:exon4:c.394C>A:p.R132S	Missense	48.4
AML_49	NPM1	NM_002520:exon11:c.861_862insTGCA:p.L287fs	InDel	50.2
AML_50	DNMT3A	NM_175629:exon17:c.1936+3G>A,p.?	Splice site	66.2
AML_50	TET2	NM_001127208:exon11:c.5633G>A:p.R1878H	Missense	40.1
AML_50	TP53	NM_000546:exon6:c.596_598del:p.199_200del	InDel	4.3
AML_50	TP53	NM_000546:exon5:c.470T>A:p.V157D	Missense	83.3
AML_51	BCOR	NM_001123385:exon10:c.4288C>T:p.Q1430X	Nonsense	45.9
AML_51	BCORL1	NM_021946:c.3268C>T:p.R1090X	Nonsense	28.3
AML_51	BCORL1	NM_021946:c.3994C>T:p.R1332X	Nonsense	10.3
AML_51	DNMT3A	NM_175629:exon23:c.2645G>A:p.R882H	Missense	50.2
AML_51	NRAS	NM_002524:exon4:c.436G>A:p.A146T	Missense	3.4
AML_51	RUNX1	NM_001754:exon4:c.325A>T:p.N109Y	Missense	48.0
AML_51	STAG2	NM_001042750:exon28:c.2857C>T:p.R953X	Nonsense	48.3
AML_52	TET2	NM_001127208:exon3:c.1294G>T:p.E432X	Nonsense	53.0
AML_52	TET2	NM_001127208:exon8:c.3987delG:p.L1329fs	InDel	52.1
AML_53	FLT3	NM_004119:exon14:c.1787_1788insTCCCCACCAGCTACAG ATGGTACAGGTGACCGGCTCCTCAGATAATGAGTACTTCT ACGTTGATTTTCAGAGA:p.E596delinsDPHQLQMVQVTGSSD NEYFYVDFRE	InDel	23.8
AML_54	CBL	NM_005188:exon8:c.1199T>G:p.M400R	Missense	79.0
AML_54	IDH1	NM_005896:exon4:c.395G>A:p.R132H	Missense	42.3
AML_54	RUNX1	NM_001754:exon6:c.611G>A:p.R204Q	Missense	8.7

Subject	Gene	Variant	Mutation type	VAF [%]
AML_54	<i>RUNX1</i>	NM_001754:exon5:c.370_371insGATG:p.V124fs	InDel	35.0
AML_54	<i>SRSF2</i>	NM_003016:exon1:c.284C>G:p.P95R	Missense	46.2
AML_55	<i>DNMT3A</i>	NM_175629:exon21:c.2409_2410insTAGG:p.P804_L805delinsX	Nonsense	53.9
AML_55	<i>RUNX1</i>	NM_001754:exon6:c.602G>A:p.R201Q	Missense	13.6
AML_55	<i>RUNX1</i>	NM_001754:exon5:c.496C>G:p.R166G	Missense	15.5
AML_55	<i>SF3B1</i>	NM_012433:exon15:c.2098A>G:p.K700E	Missense	28.3
AML_56	<i>ASXL1</i>	NM_015338:exon12:c.2925T>A:p.C975X	Nonsense	5.4
AML_56	<i>TP53</i>	NM_000546:exon6:c.659_672del:p.Y220fs	InDel	34.2
AML_57	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG:p.G642fs	InDel	23.0
AML_57	<i>BCOR</i>	NM_001123385:exon4:c.466_467insCCCCC AAAACACCGCCTGGAATAC:p.Q156fs	InDel	59.0
AML_57	<i>BCOR</i>	NM_001123385:exon4:c.464_465insCCCCC AAAACACCGCCTGGAAT:p.I155fs	InDel	5.7
AML_57	<i>RUNX1</i>	NM_001754:exon6:c.610C>T:p.R204X	Nonsense	20.5
AML_57	<i>SRSF2</i>	NM_003016:exon1:c.284C>A:p.P95H	Missense	42.5
AML_57	<i>STAG2</i>	NM_001042750:exon17:c.1535_1536AGC	InDel	15.4
AML_57	<i>STAG2</i>	NM_001042750:exon17:c.1536delA:p.A512fs	InDel	75.6
AML_58	<i>ASXL1</i>	NM_015338:exon12:c.2243delA:p.Q748fs	InDel	40.8
AML_58	<i>U2AF1</i>	NM_001025203:exon2:c.101C>T:p.S34F	Missense	47.3
AML_59	<i>ASXL1</i>	NM_015338:exon12:c.1888_1910del:p.H630fs	InDel	42.4
AML_59	<i>CBL</i>	NM_005188:exon8:c.1186T>C:p.C396R	Missense	29.0
AML_59	<i>CBL</i>	NM_005188:exon9:c.1250C>G:p.P417R	Missense	4.5
AML_59	<i>FLT3</i>	NM_004119:exon20:c.2503G>T:p.D835Y	Missense	32.0
AML_59	<i>SRSF2</i>	NM_003016:exon1:c.284C>A:p.P95H	Missense	48.5
AML_59	<i>TET2</i>	NM_001127208:exon3:c.1143delC:p.F381fs	InDel	46.2
AML_59	<i>TET2</i>	NM_001127208:exon3:c.1172delC:p.S391fs	InDel	45.7
AML_60	<i>BCORL1</i>	NM_021946:c.3304A>T:p.K1102X	Nonsense	89.1
AML_60	<i>ETV6</i>	NM_001987:exon3:c.305_306insGAAGGGGGACCTC:p.F102fs	InDel	23.1
AML_60	<i>FLT3</i>	NM_004119:exon14:c.1781_1782insGGT:p.F594delinsLV	InDel	26.7
AML_60	<i>NRAS</i>	NM_002524:exon3:c.183A>T:p.Q61H	Missense	2.5
AML_60	<i>NRAS</i>	NM_002524:exon2:c.37G>C:p.G13R	Missense	9.7
AML_61	<i>ASXL1</i>	NM_015338:exon12:c.2160delC:p.D720fs	InDel	48.4
AML_61	<i>IDH2</i>	NM_002168:exon4:c.418C>G:p.R140G	Missense	43.3
AML_61	<i>KRAS</i>	NM_033360:exon2:c.34G>C:p.G12R	Missense	3.3
AML_61	<i>NRAS</i>	NM_002524:exon2:c.35G>T:p.G12V	Missense	36.7
AML_61	<i>RUNX1</i>	NM_001754:exon7:c.694C>T:p.R232W	Missense	29.3
AML_61	<i>SRSF2</i>	NM_003016:exon1:c.284C>A:p.P95H	Missense	50.4
AML_61	<i>STAG2</i>	NM_001042750:exon20:c.1840C>T:p.R614X	Nonsense	94.7
AML_62	<i>FLT3</i>	NM_004119:exon20:c.2508_2510del:p.836_837del	InDel	2.2
AML_62	<i>IDH1</i>	NM_005896:exon4:c.395G>A:p.R132H	Missense	38.4
AML_63	<i>TP53</i>	NM_000546:exon8:c.818G>A:p.R273H	Missense	25.6
AML_63	<i>TP53</i>	NM_000546:exon6:c.657delC:p.P219fs	InDel	23.7
AML_64	<i>DNMT3A</i>	NM_175629:exon23:c.2645G>A:p.R882H	Missense	49.0
AML_64	<i>JAK2</i>	NM_004972:exon14:c.1849G>T:p.V617F	Missense	48.0
AML_64	<i>NRAS</i>	NM_002524:exon2:c.38G>T:p.G13V	Missense	48.0

Subject	Gene	Variant	Mutation type	VAF [%]
AML_64	STAG2	NM_001042750:exon6:c.328C>T:p.R110X	Nonsense	48.0
AML_64	TET2	NM_001127208:exon3:c.547dupA:p.L182fs	InDel	50.0
AML_65	ASXL1	NM_015338:exon12:c.4116_4117del:p.1372_1373del	InDel	44.7
AML_65	NRAS	NM_002524:exon2:c.38G>T:p.G13V	Missense	7.7
AML_65	RUNX1	NM_001754:exon5:c.492_493insTA:p.G165_R166delinsX	Nonsense	8.6
AML_65	SRSF2	NM_003016:exon1:c.283_284insGCC:p.P95delinsRP	InDel	47.0
AML_65	STAG2	NM_001042750:exon21:c.2096+1->T,p.?	Splice site	71.7
AML_65	TET2	NM_001127208:exon3:c.1452T>A:p.C484X	Nonsense	28.6
AML_65	TET2	NM_001127208:exon4:c.3430G>A:p.E1144K	Missense	48.9
AML_66	ASXL1	NM_015338:exon12:c.1888_1910del:p.H630fs	InDel	40.4
AML_66	IDH1	NM_005896:exon4:c.394C>G:p.R132G	Missense	45.2
AML_66	JAK2	NM_004972:exon14:c.1849G>T:p.V617F	Missense	3.4
AML_66	RUNX1	NM_001754:exon4:c.319C>T:p.R107C	Missense	44.3
AML_66	SRSF2	NM_003016:exon1:c.284C>A:p.P95H	Missense	50.8
AML_67	TP53	NM_000546:exon7:c.743G>A:p.R248Q	Missense	50.4
AML_68	FLT3	NM_004119:exon14:c.1834_1835insATGAATATGATCTCAAA TGGGAGTTTCCAAGAGAAAATTTAGAGT;p.F612delinsYEY DLKWEFPRENLEF	InDel	12.0
AML_68	FLT3	NM_004119:exon14:c.1797_1798insCTTAACGTTGATTTTCAG AGAATATGAATAT;p.D600delinsLNVDFREYEYD	InDel	4.9
AML_68	FLT3	NM_004119:exon14:c.1793_1794insCTACGTTGATTTTCAGA GAATATGA;p.E598delinsDYVDFREYE	InDel	3.9
AML_68	U2AF1	NM_001025203:exon2:c.101C>T:p.S34F	Missense	44.6
AML_69	FLT3	NM_004119:exon15:c.1837+2- >AAAGCGGGTGACCGGCTCCTCAGATAATGAGTACTTCTA CGTTGATTTTCAGAGAATATGAATATGATCTCAAATGGGAG TTTCCAAGAGAAAATTTAGAGTTTGG	Splice site	2.1
AML_69	FLT3	NM_004119:exon14:c.1815_1816insGGAAAGCGGGTGACC GGCTCCTCAGATAATGAGTACTTCTACGTTGATTTTCAGAG AATATGAATATGATCTCAAATGGGAGTTT;p.P606delinsGKR VTGSSDNEYFYVDFREYEYDLKWEFP	InDel	11.8
AML_69	FLT3	NM_004119:exon14:c.1788_1789insGATAATGAGTACTTCTA CGTTGATTTTCAGAGAA;p.Y597delinsDNEYFYVDFREY	InDel	3.6
AML_69	IDH2	NM_002168:exon4:c.419G>A:p.R140Q	Missense	41.1
AML_69	NPM1	NM_002520:exon11:c.859_860insTCTG;p.L287fs	InDel	36.8
AML_69	SRSF2	NM_003016:exon1:c.284C>T:p.P95L	Missense	29.3
AML_69	WT1	NM_024426:exon7:c.1195A>C:p.N399H	Missense	47.4
AML_70	BCOR	NM_001123385:exon4:c.524_527del:p.175_176del	InDel	65.3
AML_70	NRAS	NM_002524:exon3:c.180_181insGCTTCC:p.Q61delinsASQ	InDel	15.3
AML_70	RUNX1	NM_001754:exon5:c.495_496insTT:p.R166fs	InDel	43.3
AML_70	U2AF1	NM_001025203:exon2:c.101C>A:p.S34Y	Missense	41.0
AML_71	IDH2	NM_002168:exon4:c.515G>A:p.R172K	Missense	35.0
AML_72	BCOR	NM_001123385:exon11:c.4428+1G>A,p.?	Splice site	36.2
AML_72	DNMT3A	NM_175629:exon23:c.2728G>C:p.A910P	Missense	38.2
AML_72	DNMT3A	NM_175629:exon8:c.942G>A:p.W314X	Nonsense	45.8
AML_72	IDH2	NM_002168:exon4:c.419G>A:p.R140Q	Missense	40.2
AML_73	FLT3	NM_004119:exon20:c.2503G>T:p.D835Y	Missense	3.8
AML_73	NPM1	NM_002520:exon11:c.859_860insTCTG;p.L287fs	InDel	37.0
AML_73	NRAS	NM_002524:exon2:c.38G>A:p.G13D	Missense	14.4

Subject	Gene	Variant	Mutation type	VAF [%]
AML_73	<i>PTPN11</i>	NM_002834:exon3:c.179G>T:p.G60V	Missense	3.3
AML_73	<i>RAD21</i>	NM_006265:exon8:c.933_934insTA:p.T312_V313delinsX	Nonsense	33.0
AML_74	<i>FLT3</i>	NM_004119:exon15:c.1837+1->ATTTTCAGAGAATATGAATATGATCTCAAATGGGAGTTTCC AAGAGAAAATTTAGAGTTTG	Splice site	7.5
AML_74	<i>FLT3</i>	NM_004119:exon14:c.1778_1779insCCAGCTACAGATGGTA CAGGTGACCGGCTCCTCAGATAATGAGTACTTCTACGTTG A:p.D593delinsDQLQMVQVTGSSDNEYFYVD	InDel	33.2
AML_74	<i>IDH2</i>	NM_002168:exon4:c.419G>A:p.R140Q	Missense	41.5
AML_75	<i>ASXL1</i>	NM_015338:exon12:c.1773C>A:p.Y591X	Nonsense	23.6
AML_75	<i>EZH2</i>	NM_004456:exon11:c.1262_1271del:p.T421fs	InDel	52.3
AML_75	<i>IDH1</i>	NM_005896:exon4:c.395G>A:p.R132H	Missense	34.5
AML_75	<i>PTPN11</i>	NM_002834:exon3:c.188A>G:p.Y63C	Missense	29.2
AML_75	<i>RUNX1</i>	NM_001754:exon9:c.1003_1013del:p.Q335fs	InDel	12.7
AML_75	<i>RUNX1</i>	NM_001754:exon6:c.508+1G>T,p.?	Splice site	15.7
AML_76	<i>DNMT3A</i>	NM_175629:exon23:c.2645G>A:p.R882H	Missense	35.5
AML_76	<i>KRAS</i>	NM_033360:exon2:c.35G>A:p.G12D	Missense	33.6
AML_76	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	38.3
AML_77	<i>BCOR</i>	NM_001123385:exon8:c.3649C>T:p.R1217X	Nonsense	16.3
AML_77	<i>DNMT3A</i>	NM_175629:exon8:c.912delC:p.S304fs	InDel	28.3
AML_77	<i>IDH2</i>	NM_002168:exon4:c.419G>A:p.R140Q	Missense	23.4
AML_78	<i>BCOR</i>	NM_001123385:exon10:c.4174_4225del:p.V1392fs	InDel	4.1
AML_78	<i>BCORL1</i>	NM_021946:c.2843_2844del:p.Q948fs	InDel	86.6
AML_78	<i>NRAS</i>	NM_002524:exon2:c.38G>A:p.G13D	Missense	42.9
AML_78	<i>RUNX1</i>	NM_001754:exon9:c.1290dupG:p.P431fs	InDel	43.2
AML_78	<i>U2AF1</i>	NM_001025203:exon2:c.101C>T:p.S34F	Missense	47.8
AML_79	<i>DDX41</i>	NM_016222:exon15:c.1586_1587del:p.T529fs	InDel	46.8
AML_79	<i>DNMT3A</i>	NM_175629:exon13:c.1502A>G:p.N501S	Missense	54.1
AML_79	<i>EZH2</i>	NM_004456:exon17:c.1973_1974del:p.R658fs	InDel	60.5
AML_80	<i>ASXL1</i>	NM_015338:exon12:c.2084dupA:p.Q695fs	InDel	39.1
AML_80	<i>RUNX1</i>	NM_001754:exon4:c.230dupG:p.S77fs	InDel	45.6
AML_80	<i>SRSF2</i>	NM_003016:exon1:c.284C>A:p.P95H	Missense	40.6
AML_80	<i>STAG2</i>	NM_001042750:exon27:c.2694_2695insTGGTAGAGTTGA:p.D898delinsDWX	Nonsense	29.4
AML_81	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG:p.G642fs	InDel	41.4
AML_81	<i>EZH2</i>	NM_004456:exon18:c.2050C>T:p.R684C	Missense	9.9
AML_81	<i>NRAS</i>	NM_002524:exon2:c.38G>A:p.G13D	Missense	42.1
AML_81	<i>RUNX1</i>	NM_001754:exon6:c.553C>A:p.Q185K	Missense	42.3
AML_81	<i>SRSF2</i>	NM_003016:exon1:c.284C>A:p.P95H	Missense	50.0
AML_81	<i>STAG2</i>	NM_001042750:exon30:c.3097C>T:p.R1033X	Nonsense	73.5
AML_82	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG:p.G642fs	InDel	15.4
AML_82	<i>PHF6</i>	NM_032458:exon6:c.442G>T:p.E148X	Nonsense	32.9
AML_82	<i>U2AF1</i>	NM_001025203:exon2:c.101C>T:p.S34F	Missense	30.1
AML_83	<i>EZH2</i>	NM_004456:exon19:c.2161A>G:p.I721V	Missense	54.0
AML_83	<i>FLT3</i>	NM_004119:exon20:c.2503G>T:p.D835Y	Missense	13.4
AML_83	<i>FLT3</i>	NM_004119:exon14:c.1796_1796delinsCCCTAGATAATGAG TACTTCTACGTTGATTTTCAGAGAATATGAAT	InDel	0.5

Subject	Gene	Variant	Mutation type	VAF [%]
AML_83	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG;p.L287fs	InDel	36.3
AML_83	<i>PTPN11</i>	NM_002834:exon3:c.179G>C;p.G60A	Missense	6.8
AML_83	<i>WT1</i>	NM_024426:exon7:c.1142C>A;p.S381X	Nonsense	17.2
AML_83	<i>WT1</i>	NM_024426:exon1:c.252_256del;p.84_86del	InDel	33.3
AML_84	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG;p.G642fs	InDel	27.0
AML_84	<i>BCOR</i>	NM_001123385:exon11:c.4455_4456insGG;p.K1486fs	InDel	80.9
AML_84	<i>BCOR</i>	NM_001123385:exon11:c.4455_4455delinsTGG	InDel	86.9
AML_84	<i>EZH2</i>	NM_004456:exon18:c.2051G>A;p.R684H	Missense	5.2
AML_84	<i>HNRNPK</i>	NM_002140:exon16:c.1192-2A>G,p.?	Splice site	40.6
AML_84	<i>STAG2</i>	NM_001042750:exon19:c.1810C>T;p.R604X	Nonsense	88.2
AML_85	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG;p.G642fs	InDel	23.6
AML_85	<i>GATA1</i>	NM_002049:exon3:c.325G>C;p.V109L	Missense	92.5
AML_85	<i>IDH2</i>	NM_002168:exon4:c.419G>A;p.R140Q	Missense	46.2
AML_85	<i>SRSF2</i>	NM_003016:exon1:c.284C>A;p.P95H	Missense	48.8
AML_85	<i>STAG2</i>	NM_001042750:exon20:c.1907dupA;p.Y636_H637delinsX	Nonsense	91.9
AML_86	<i>DNMT3A</i>	NM_175629:exon20:c.2322+1G>A,p.?	Splice site	37.7
AML_86	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG;p.L287fs	InDel	51.5
AML_86	<i>NRAS</i>	NM_002524:exon2:c.34G>A;p.G12S	Missense	4.1
AML_86	<i>PTPN11</i>	NM_002834:exon3:c.179G>T;p.G60V	Missense	36.9
AML_86	<i>TET2</i>	NM_001127208:exon6:c.3707T>A;p.I1236N	Missense	2.7
AML_87	<i>KRAS</i>	NM_033360:exon2:c.35G>T;p.G12V	Missense	5.1
AML_87	<i>NRAS</i>	NM_002524:exon2:c.35G>A;p.G12D	Missense	6.8
AML_87	<i>TET2</i>	NM_001127208:exon11:c.4883_4884insTCCATCATATCAATGCAATGGAAACC;p.Y1628fs	InDel	13.9
AML_88	<i>DNMT3A</i>	NM_175629:exon14:c.1627G>T;p.G543C	Missense	12.1
AML_88	<i>FLT3</i>	NM_004119:exon15:c.1837+10>AGAATAATGAGTACTTCTACGTTGATTTCAGAGAATATGAATATGATCTCAAATGGGAGTTTCCAAGAGAAAATTTAGAGTTTG	InDel	1.6
AML_88	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG;p.L287fs	InDel	20.4
AML_88	<i>WT1</i>	NM_024426:exon7:c.1144_1145insTCGG;p.A382fs	InDel	6.2
AML_89	<i>ASXL1</i>	NM_015338:exon12:c.2077C>T;p.R693X	Nonsense	8.3
AML_89	<i>FLT3</i>	NM_004119:exon14:c.1808_1808delinsTCAGAGAATATGAATATGATCTCAAAT	InDel	0.3
AML_89	<i>NRAS</i>	NM_002524:exon3:c.182A>C;p.Q61P	Missense	4.1
AML_89	<i>NRAS</i>	NM_002524:exon2:c.38G>T;p.G13V	Missense	39.1
AML_89	<i>RUNX1</i>	NM_001754:exon5:c.374delC;p.P125fs	InDel	46.0
AML_89	<i>SRSF2</i>	NM_003016:exon1:c.284C>A;p.P95H	Missense	44.6
AML_89	<i>STAG2</i>	NM_001042750:exon9:c.687dupT;p.A229fs	InDel	94.9
AML_89	<i>TET2</i>	NM_001127208:exon11:c.5072_5073insTA;p.S1691fs	InDel	46.9
AML_90	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG;p.G642fs	InDel	19.5
AML_90	<i>TET2</i>	NM_001127208:exon10:c.4210C>T;p.R1404X	Nonsense	17.1
AML_91	<i>JAK2</i>	NM_004972:exon14:c.1849G>T;p.V617F	Missense	98.6
AML_92	<i>DNMT3A</i>	NM_175629:exon16:c.1898C>A;p.P633H	Missense	47.4
AML_92	<i>IDH2</i>	NM_002168:exon4:c.419G>A;p.R140Q	Missense	6.4
AML_92	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG;p.L287fs	InDel	43.1
AML_92	<i>NRAS</i>	NM_002524:exon2:c.38G>A;p.G13D	Missense	4.3

Subject	Gene	Variant	Mutation type	VAF [%]
AML_92	<i>NRAS</i>	NM_002524:exon2:c.35G>A:p.G12D	Missense	37.1
AML_92	<i>RAD21</i>	NM_006265:exon13:c.1621-1G>T,p.?	Splice site	39.9
AML_93	<i>DNMT3A</i>	NM_175629:exon19:c.2311C>T:p.R771X	Nonsense	51.0
AML_93	<i>DNMT3A</i>	NM_175629:exon15:c.1687G>A:p.V563M	Missense	52.1
AML_93	<i>IDH2</i>	NM_002168:exon4:c.419G>A:p.R140Q	Missense	48.2
AML_93	<i>JAK2</i>	NM_004972:exon14:c.1849G>T:p.V617F	Missense	47.6
AML_93	<i>SRSF2</i>	NM_003016:exon1:c.284C>A:p.P95H	Missense	40.1
AML_94	<i>ASXL1</i>	NM_015338:exon12:c.1888_1910del:p.630_637del	InDel	21.5
AML_94	<i>ASXL1</i>	NM_015338:exon12:c.2118_2119insAC:p.H706fs	InDel	8.9
AML_94	<i>DNMT3A</i>	NM_175629:exon19:c.2255_2257del:p.752_753del	InDel	47.5
AML_94	<i>DNMT3A</i>	NM_175629:exon19:c.2206C>T:p.R736C	Missense	47.6
AML_94	<i>IDH2</i>	NM_002168:exon4:c.515G>A:p.R172K	Missense	45.4
AML_95	<i>NRAS</i>	NM_002524:exon3:c.182A>G:p.Q61R	Missense	11.2
AML_95	<i>TP53</i>	NM_000546:exon8:c.844C>T:p.R282W	Missense	63.0
AML_95	<i>TP53</i>	NM_000546:exon4:c.97-2A>G,p.?	Splice site	20.0
AML_96	<i>DNMT3A</i>	NM_175629:exon23:c.2645G>A:p.R882H	Missense	38.5
AML_96	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	36.5
AML_96	<i>NRAS</i>	NM_002524:exon2:c.35G>A:p.G12D	Missense	37.9
AML_96	<i>U2AF1</i>	NM_001025203:exon2:c.101C>T:p.S34F	Missense	41.6
AML_97	<i>DNMT3A</i>	NM_175629:exon23:c.2711C>T:p.P904L	Missense	18.7
AML_97	<i>IDH2</i>	NM_002168:exon4:c.419G>A:p.R140Q	Missense	19.6
AML_97	<i>KRAS</i>	NM_033360:exon2:c.35G>A:p.G12D	Missense	2.9
AML_98	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	31.0
AML_98	<i>TET2</i>	NM_001127208:exon3:c.1441C>T:p.Q481X	Nonsense	34.1
AML_98	<i>TET2</i>	NM_001127208:exon3:c.2491C>T:p.Q831X	Nonsense	56.3
AML_99	<i>DNMT3A</i>	NM_175629:exon16:c.1906G>T:p.V636L	Missense	96.3
AML_99	<i>FLT3</i>	NM_004119:exon14:c.1810_1811insTAGTTGATTCAGAGAA TATGAATATGATCTCAAATGGG;p.E604delinsVDFREYEYD LKWE	InDel	29.4
AML_99	<i>FLT3</i>	NM_004119:exon14:c.1792_1793insTCGATTCAGAGAATAT G;p.E598delinsVDFREYE	InDel	12.7
AML_99	<i>RUNX1</i>	NM_001754:exon4:c.330_331insT:p.T111fs	InDel	22.8
AML_100	<i>ETV6</i>	NM_001987:exon6:c.1138T>G:p.W380G	Missense	25.0
AML_100	<i>KRAS</i>	NM_033360:exon3:c.183A>C:p.Q61H	Missense	14.9
AML_100	<i>WT1</i>	NM_024426:exon9:c.1395C>A:p.H465Q	Missense	5.4
AML_100	<i>WT1</i>	NM_024426:exon7:c.1199dupA:p.K400fs	InDel	3.1
AML_101	<i>ASXL1</i>	NM_015338:exon12:c.1927dupG:p.G642fs	InDel	23.2
AML_101	<i>KDM6A</i>	NM_021140:exon13:c.1310C>T:p.A437V	Missense	2.5
AML_101	<i>RUNX1</i>	NM_001754:exon9:c.989dupT:p.F330fs	InDel	39.0
AML_101	<i>TET2</i>	NM_001127208:exon7:c.3866G>T:p.C1289F	Missense	36.4
AML_101	<i>U2AF1</i>	NM_001025203:exon6:c.467G>A:p.R156H	Missense	25.5
AML_102	<i>BCOR</i>	NM_001123385:exon13:c.4742-1G>T,p.?	Splice site	25.0
AML_102	<i>NRAS</i>	NM_002524:exon2:c.34G>T:p.G12C	Missense	13.5
AML_102	<i>RUNX1</i>	NM_001754:exon6:c.509-3C>G,p.?	Splice site	58.8
AML_102	<i>SF3B1</i>	NM_012433:exon15:c.2098A>G:p.K700E	Missense	52.4
AML_102	<i>TET2</i>	NM_001127208:exon6:c.3641G>A:p.R1214Q	Missense	20.0

Subject	Gene	Variant	Mutation type	VAF [%]
AML_103	ASXL1	NM_015338:exon12:c.1931_1932insGGGT:p.G644fs	InDel	23.9
AML_103	RUNX1	NM_001754:exon5:c.496C>T:p.R166X	Nonsense	42.1
AML_103	SRSF2	NM_003016:exon1:c.284C>A:p.P95H	Missense	39.5
AML_103	TET2	NM_001127208:exon3:c.1937_1938insAGTGG:p.T646fs	InDel	28.0
AML_103	TET2	NM_001127208:exon6:c.3782G>A:p.R1261H	Missense	23.7
AML_104	RUNX1	NM_001754:exon6:c.508+1G>A,p.?	Splice site	36.3
AML_104	RUNX1	NM_001754:exon5:c.508G>C:p.G170R	Missense	26.8
AML_104	RUNX1	NM_001754:exon5:c.422C>A:p.S141X	Nonsense	4.3
AML_104	SF3B1	NM_012433:exon15:c.2098A>G:p.K700E	Missense	40.8
AML_105	DNMT3A	NM_175629:exon23:c.2645G>A:p.R882H	Missense	43.6
AML_105	KRAS	NM_033360:exon3:c.176C>G:p.A59G	Missense	4.7
AML_105	NPM1	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	47.0
AML_105	PTPN11	NM_002834:exon3:c.182A>G:p.D61G	Missense	27.5
AML_105	PTPN11	NM_002834:exon3:c.213T>A:p.F71L	Missense	6.0
AML_106	PTPN11	NM_002834:exon3:c.181G>T:p.D61Y	Missense	10.2
AML_106	TP53	NM_000546:exon6:c.646G>A:p.V216M	Missense	82.6
AML_107	NPM1	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	20.2
AML_107	NRAS	NM_002524:exon2:c.35G>A:p.G12D	Missense	3.3
AML_107	SRSF2	NM_003016:exon1:c.284C>T:p.P95L	Missense	25.6
AML_107	TET2	NM_001127208:exon7:c.3852_3854del:p.1284_1285del	InDel	37.7
AML_107	TET2	NM_001127208:exon10:c.4393C>T:p.R1465X	Nonsense	46.1
AML_108	DNMT3A	NM_175629:exon23:c.2678G>C:p.W893S	Missense	39.4
AML_108	FLT3	NM_004119:exon14:c.1831_1832insTTGATTTTCAGAGAATATGAATATGATCTCAAATGGGAGTTTCCAAGAGAAAATTTAG:p.E611delinsVDFREYEDLKWEFPRENLE	InDel	22.8
AML_108	NPM1	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	46.3
AML_109	DNMT3A	NM_175629:exon23:c.2645G>A:p.R882H	Missense	44.0
AML_109	FLT3	NM_004119:exon14:c.1816_1817insAAAATGAGTACTTCTACGTTGATTTTCAGAGAATATGAATATGATCTCAAATGGGAGTTTC:p.P606delinsQNEYFYVDFREYEDLKWEFP	InDel	5.6
AML_109	FLT3	NM_004119:exon14:c.1793_1794insCTACGTTGATTTTCAGAGAATATGA:p.E598delinsDYVDFREYE	InDel	3.0
AML_109	FLT3	NM_004119:exon14:c.1784_1785insCCCTTATTTTCAG:p.R595delinsSPYFR	InDel	2.9
AML_109	FLT3	NM_004119:exon14:c.1783_1784insCCTCAGATAATGAGTACTTCTACGTTGATTTCA:p.R595delinsTSDNEYFYVDFR	InDel	1.5
AML_109	KRAS	NM_033360:exon2:c.35G>A:p.G12D	Missense	7.5
AML_109	NPM1	NM_002520:exon11:c.859_860insTCTG:p.L287fs	InDel	39.3
AML_109	NRAS	NM_002524:exon2:c.38G>A:p.G13D	Missense	6.1
AML_109	PTPN11	NM_002834:exon3:c.205G>A:p.E69K	Missense	3.9
AML_109	WT1	NM_024426:exon7:c.1140_1141insCCGG:p.S381fs	InDel	6.4
AML_110	ASXL1	NM_015338:exon12:c.1927dupG:p.G642fs	InDel	21.0
AML_110	PTPN11	NM_002834:exon13:c.1529A>T:p.Q510L	Missense	23.2
AML_110	RUNX1	NM_001754:exon7:c.720_733del:p.240_245del	InDel	53.0
AML_110	SRSF2	NM_003016:exon1:c.284C>T:p.P95L	Missense	17.4
AML_110	STAG2	NM_001042750:exon5:c.265dupA:p.V88fs	InDel	37.8
AML_110	STAG2	NM_001042750:exon5:c.269_269delinsAG	InDel	28.3
AML_110	STAG2	NM_001042750:exon5:c.269T>G:p.M90R	Missense	38.5

Subject	Gene	Variant	Mutation type	VAF [%]
AML_111	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG;p.L287fs	InDel	37.8
AML_111	<i>PTPN11</i>	NM_002834:exon3:c.179G>T;p.G60V	Missense	44.2
AML_111	<i>STAG2</i>	NM_001042750:exon17:c.1535-3T>A,p.?	Splice site	47.8
AML_112	<i>BCOR</i>	NM_001123385:exon4:c.1555_1559del;p.E519fs	InDel	80.7
AML_112	<i>FLT3</i>	NM_004119:exon14:c.1794_1795insAAGGTGACCGGCTCCTCAGATAATGAGTACTTCTACGTTGATTTTCAGAGAATATGAA:p.Y599delinsKVTGSSDNEYFYVDFREYEY	InDel	16.5
AML_112	<i>NRAS</i>	NM_002524:exon3:c.176C>A;p.A59D	Missense	14.1
AML_112	<i>STAG2</i>	NM_001042750:exon9:c.705_706insT;p.L235fs	InDel	79.1
AML_112	<i>STAG2</i>	NM_001042750:exon9:c.706_706delinsTG	InDel	8.9
AML_112	<i>U2AF1</i>	NM_001025203:exon2:c.101C>T;p.S34F	Missense	42.8
AML_113	<i>ASXL1</i>	NM_015338:exon12:c.1888_1910del;p.H630fs	InDel	39.5
AML_113	<i>KRAS</i>	NM_033360:exon2:c.64C>A;p.Q22K	Missense	10.9
AML_113	<i>SRSF2</i>	NM_003016:exon1:c.284C>T;p.P95L	Missense	20.6
AML_113	<i>TET2</i>	NM_001127208:exon3:c.1110T>A;p.Y370X	Nonsense	68.4
AML_114	<i>DNMT3A</i>	NM_175629:exon23:c.2645G>A;p.R882H	Missense	48.0
AML_114	<i>FLT3</i>	NM_004119:exon14:c.1793_1794insCTTCGAAAGCCAGCTACAGATGGTACAGGTGACCGGCTCCTCAGATAATGAGTACTTCTACGTTGATTTTCAGAGAATATGA:p.E598delinsDFESQLQMVQVTGSSDNEYFYVDFREYE	InDel	9.0
AML_114	<i>NPM1</i>	NM_002520:exon11:c.859_860insTCTG;p.L287fs	InDel	43.0
AML_114	<i>WT1</i>	NM_024426:exon7:c.1142_1143insAACAAGAGTC;p.S381fs	InDel	25.0
AML_115	<i>ASXL1</i>	NM_015338:exon12:c.2077C>T;p.R693X	Nonsense	16.0
AML_115	<i>CEBPA</i>	NM_004364:exon1:c.961_962insC;p.N321fs	InDel	65.0
AML_115	<i>CEBPA</i>	NM_004364:exon1:c.178dupA;p.T60fs	InDel	34.0
AML_115	<i>SRSF2</i>	NM_003016:exon1:c.284C>T;p.P95L	Missense	19.0
AML_115	<i>STAG2</i>	NM_001042750:exon10:c.891C>A;p.Y297X	Nonsense	94.0
AML_116	<i>DNMT3A</i>	NM_175629:exon19:c.2196T>G;p.F732L	Missense	43.6
AML_116	<i>FLT3</i>	NM_004119:exon14:c.1808_1809insTCCAGATAATGAGTACTTCTACGTTGATTTTCAGAGAATATGAATATGATCTCAAATG:p.W603delinsCPDNEYFYVDFREYEYDLKW	InDel	27.7
AML_116	<i>NPM1</i>	NM_002520:exon11:c.860_861insCTGC;p.L287fs	InDel	33.6
AML_116	<i>WT1</i>	NM_024426:exon7:c.1142C>A;p.S381X	Nonsense	46.1
AML_117	<i>IDH1</i>	NM_005896:exon4:c.394C>T;p.R132C	Missense	18.6

Abbreviations: VAF, variant allele frequency; InDel, insertion/deletion variant; MDS, myelodysplastic neoplasm; AML, acute myeloid leukemia.

Supplemental Table 12: Summary statistics of mutations identified in individuals undergoing total hip arthroplasty and patients with MDS or sAML.

	THA (n=261)	MDS (n=91)	sAML (n=123)
Individuals with ≥ 1 somatic variant	127/261 (49%)	84/91 (92%)	117/123 (95%)
Variants per individual (range)	0 - 5	0 - 9	0 - 11
Among individuals with ≥ 1 variant:			
- Median number of variants	1	3	4
- VAF [%] (median, range)	2.7 (1 - 44)	19 (1 - 87)	37 (0.3 - 99)

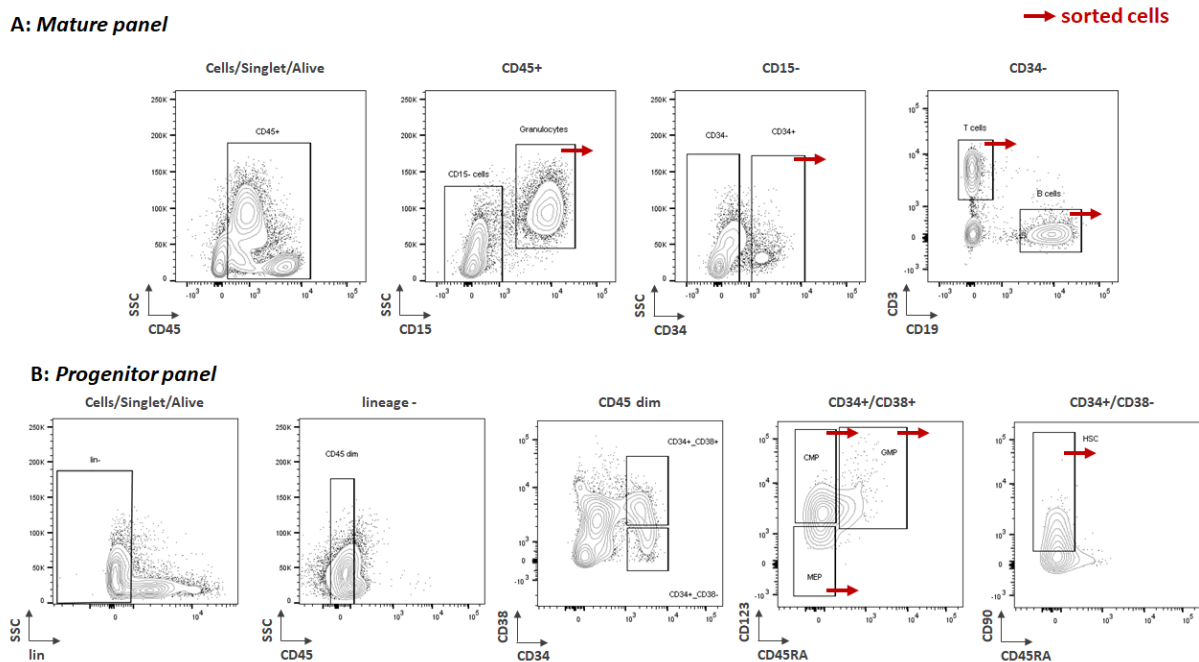
Abbreviations: VAF, variant allele frequency; THA, total hip arthroplasty; MDS, myelodysplastic neoplasm; AML, acute myeloid leukemia.

Supplemental Table 13: Comparison of *DNMT3A*, *TET2* and *ASXL1* variant properties between CH, MDS and sAML.

	CH (n=127)	MDS (n=91)	sAML (n=123)
<i>DNMT3A</i> p.R882 vs. others	4/87 (5%)	9/28 (32%)	14/37 (38%)
<i>TET2</i> InDel or nonsense vs. others	27/47 (57%)	43/62 (69%)	27/37 (73%)
<i>ASXL1</i> c.1934dup vs. others	3/12 (25%)	9/22 (41%)	12/34 (35%)

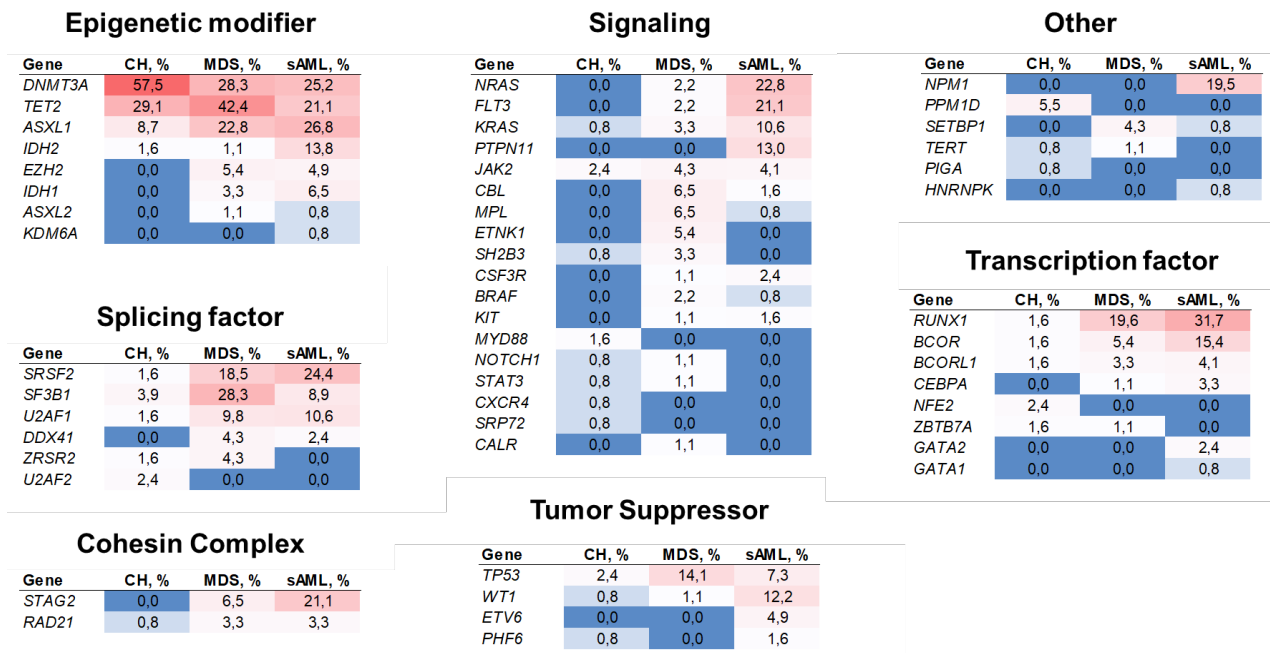
Abbreviations: InDel, insertion/deletion variant; MDS, myelodysplastic neoplasm; AML, acute myeloid leukemia.

Supplemental Figures



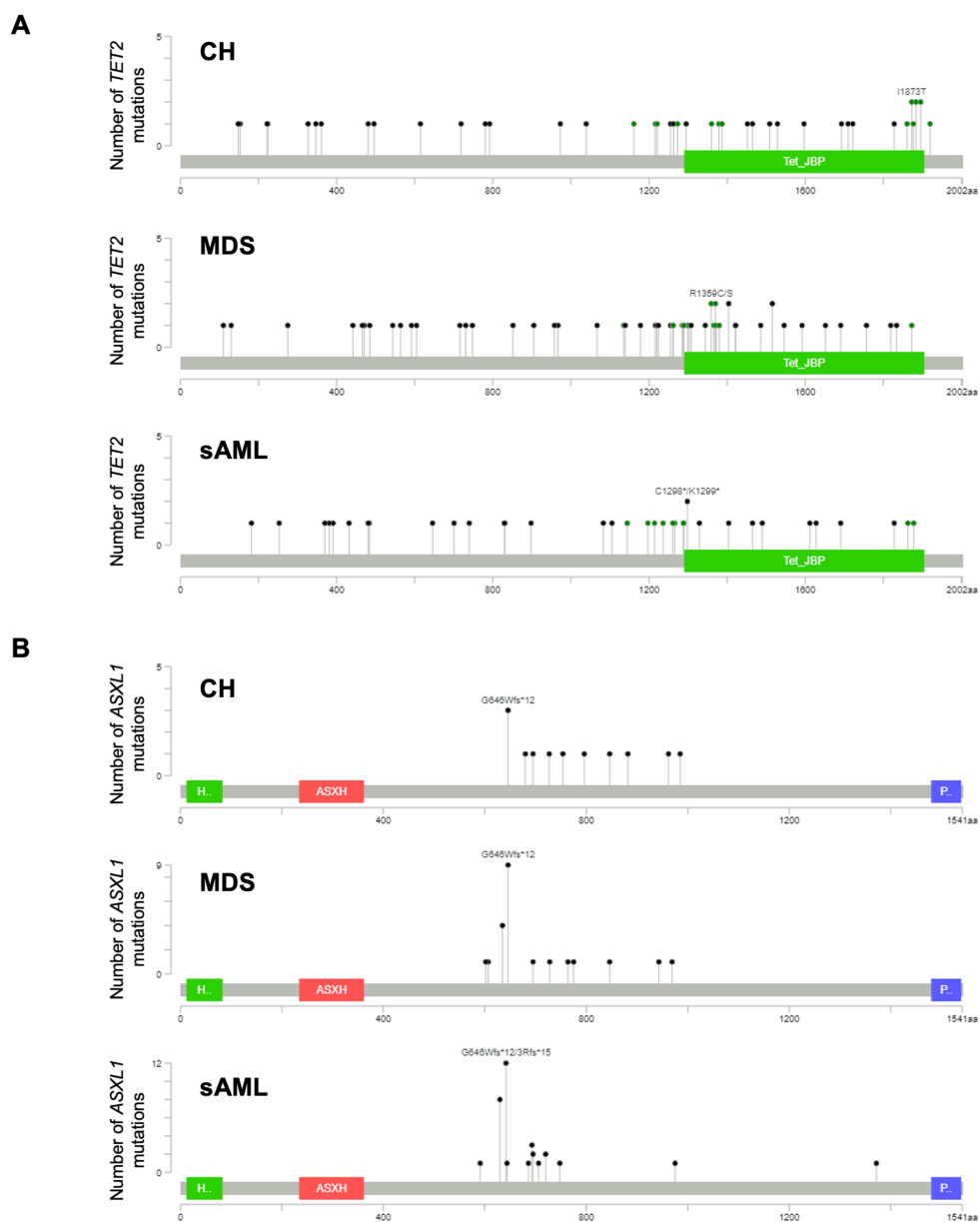
Supplemental Figure 1: Gating strategy for sorting BM subpopulations from individuals with CH carrying *ASXL1* mutations.

A: After dead cell exclusion, CD15⁺ (granulocytes) cells were sorted, the negative fraction was then used to sort CD34⁺ cells and the subsequent CD15⁻CD34⁻ fraction was used to sort CD3⁺ (T-cells) and CD19⁺ (B-cells). B: After dead cell exclusion, lineage negative cells (CD4⁻CD8a⁻CD15⁻CD19⁻CD235a⁻) were selected and subsequently the CD45dim population. The CD34⁺CD38⁺ subpopulation was used to sort for CD45RA⁻CD123⁺ (CMPs), CD45RA⁺CD123⁺ (GMPs) and CD45⁻CD123⁻ (MEPs), while the CD34⁺CD38⁻ subpopulation was used to sort for CD45RA⁻CD90⁺ (HSCs).



Supplemental Figure 2: Frequency of mutations in individuals with CH, MDS or sAML, grouped according to functional category

Classification of genetic variants into functional categories was performed according to Döhner et al., N Engl J Med 2015; 373:1136–1152.



Supplemental Figure 3: Intra-genic localization of variants in CH, MDS and sAML.

(A) *TET2* variants; (B) *ASXL1* variants. Green dots indicate missense mutations, black dots indicate InDels, brown dots indicate nonsense mutations. Plots were generated using the MutationMapper tool (Gao et al., *Sci. Signal.* 2013 and Cerami et al., *Cancer Discov.* 2012).